Dermatology diseases Index

FIRST INDEX OF SKIN DISEASES IN THE WORLD

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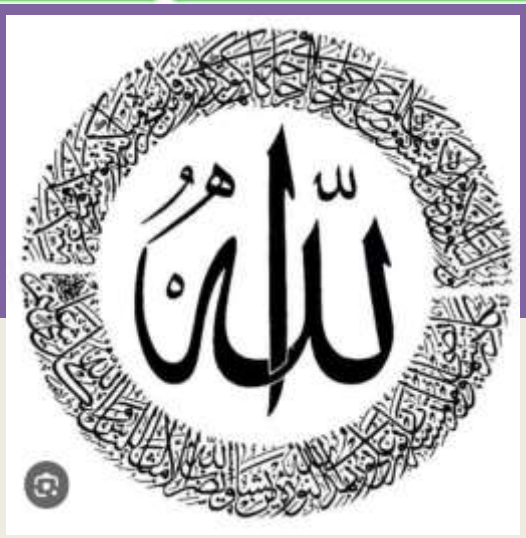
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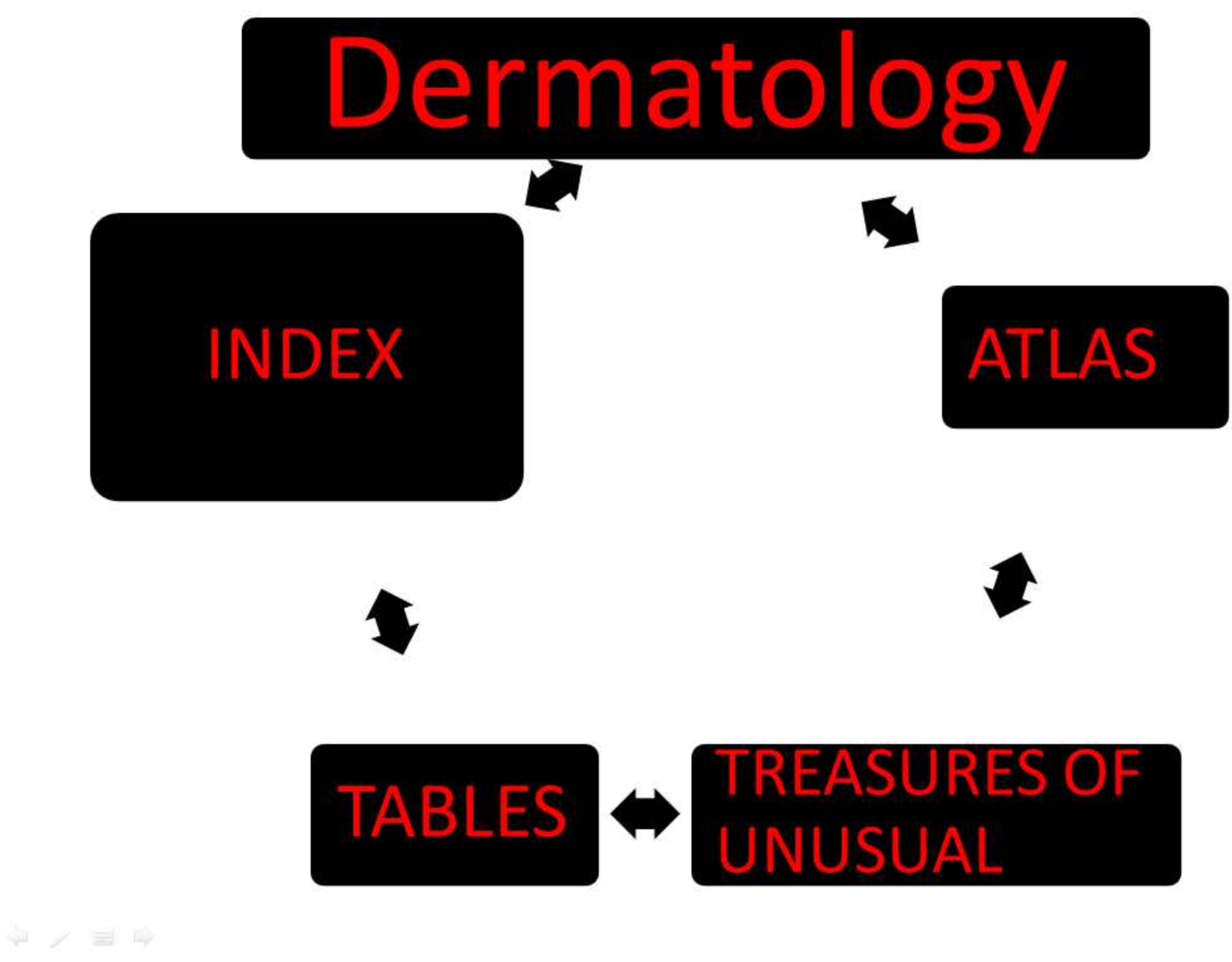
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Your gate to FIRST and LARGEST index of skin diseases

Here

Author 's words

Hello everyone and every doctor to my index

This index represents the only largest list and index of skin diseases evermade till now in the literature.

I put between your hands treasure of names of skin diseases , updated almost daily from new issues of international journals.

I hope my index is beneficial to you

Kind regards,

Author:Hany Salah Soliman

Dermatology specialist-Egypt

NOTE

You can refer to the book of collection for further details about signs

29 APRIL 2026

(1) Nail diseases

1. Absent lunulae =Anolunula
2. Acro osteolysis
3. Acropathie ulcéro mutilante = Acropathia ulcero-mutilans
4. Alopecia areata: nail signs
5. Amelanotic melanoma of nail
6. Aneurysmal bone cyst
7. Angioleiomyoma
8. Anonychia
9. Anonychia–onychodystrophy with brachydactyly type b and ectrodactyly
10. Anonychia with bizarre flexural pigmentation
11. Asymmetric gait nail unit syndrome
12. Basal cell carcinoma
13. Beau's lines
14. Bissell's line
15. Bidet nail
16. Bifid thumb
17. Blue /Azure lunulae
18. Bowen disease of nail
19. Botryomycoma
20. Brachyonychia (short nails)
21. Brittle nail
22. Brittle nail syndrome
23. Bullous diseases :nail signs
24. Carpal tunnel syndrome
25. Chevron nail
26. Chromonychia
27. Circumferential nail
28. Cirsoid angioma
29. Claw-like nail

30. Clubbing of nails
31. Coccal nail-fold angiomatosis
32. Congenital hypertrophy of the lateral fold of the hallux
33. Crumbling of nail
34. Curly fingernails
35. Curved nail of the fourth toe
36. Darier disease: nail signs
37. Dermatofibrosarcoma protuberans of nail
38. Dermatomyositis nail signs
39. Disappearing digit
40. Distal nail embedding
41. Distal subungual keratosis
42. Dolichonychia (long nails)
43. Double nail anomaly
44. Drug-Induced Nail Abnormalities
45. Dyskeratosis congenita: nail
46. Dystelephalangy= Kirner deformity
47. Ectopic nail =onychoheterotopia
48. Ectrodactyly
49. Eczema : nail signs
50. Elkonyxis
51. Erythronychia
52. Familial nail dysplasia
53. Familial nail dystrophy
54. Familial twenty-nail dystrophy
55. Fibrokeratoma of nail
56. Fibroma
57. Fibromyxoma
58. Subungual filamentous tumor
59. Finger sucking
60. Frictional melanonychia
61. Glomus tumour
62. Graft vs host disease GVHD :nail signs

63. **Granular cell tumor**
64. **Grasp reflex multiple ingrown fingernails**
65. **Green nail syndrome =Goldman-Fox syndrome**
66. **Habit tic deformity**
67. **Half and half nail**
68. **Hallux**
69. **Hangnail**
70. **Haplonychia**
71. **Harpoon nail**
72. **Heloma**
73. **Hemionychophagi**
74. **Herpes simplex :nail**
75. **Hooked nail**
76. **Hyperonychia**
77. **Hyponychia**
78. **Nail Hypoplasia**
79. **Idiopathic polydactylous longitudinal erythronychia**
80. **Ingrown toe nail= onychocryptosis**
81. **involuted toenai**
82. **Iso kikuchi syndrome=congenital onychodysplasia of index finger COIF**
83. **Kaposi sarcoma**
84. **Keratoacanthoma**
85. **Keratin granulations**
86. **Koilonychia (spoon nail)**
87. **Koenen's periungual fibromas**
88. **Lacquer nail**
89. **Leprosy nail signs**
90. **True leukonychia: punctate & striate & diffuse leukonychia (porcelain nails & leukonychia totalis)**
91. **Leukonychia variegata**
92. **Apparent leukonychia**

93. **Leukonychia totalis with epiphyseal dysplasia (Lowry–Wood) syndrome**
94. **Leukonychia totalis with PPK and congenital alopecia**
95. **Leukonychia with pili torti**
96. **Leukonychia, axonal neuropathy, myopathic features, dilated cardiomyopathy, conduction disturbances and arrhythmia**
97. **Leukonychia, duodenal ulcer, and gallstones**
98. **Leukonychia, onychorrhexis, hypoparathyroidism, dental changes, cataract syndrome**
99. **Leukonychia with koilonychias**
100. **Striated leukonychia with eruptive milia**
101. **Lichen planus : nail signs**
102. **Longitudinal furrows**
103. **Longitudinal grooves**
104. **Longitudinal ridges**
105. **Macronychia**
106. **Malalignment of the great toenails**
107. **Median canaliform dystrophy of heller**
108. **Mee's line**
109. **Megalodactyly**
110. **Melanonychia**
111. **Microlunula**
112. **Micronychia**
113. **Micronychia with trisomy 21**
114. **Muehrcke's line**
115. **Myxoid cyst= Myxoid pseudocysts of the digits**
116. **Nail apparatus melanocytic naevus**
117. **Nail apparatus melanoma**
118. **nail degloving syndrome**
119. **Nail apparatus lentigo**
120. **Nail beads**
121. **Nail biting**
122. **Nail degloving syndrome**

123. Nail dystrophy
124. Nail dystrophy–deafness syndrome= robinson's syndrome
125. Nail fragility
126. Nail fragility syndrome
127. Nail patella syndrome = Hereditary Onycho-Osteodysplasia= Fong Disease
128. Nail ridges
129. Neapolitan nail
130. Oblique lines=chevron nail=herringbone nail
131. Oil spot
132. Onychauxis
133. Onychoatrophy
134. Onychoblastoma
135. Onychocryptosis
136. Onychocytic carcinoma
137. Onychocytic matricoma
138. Onychodaknomania
139. Onychodystrophy
140. Onychogryphosis= ram's horn nail
141. Onychoheterotopia
142. Onycholemmal carcinoma
143. Onycholemmal cysts
144. Onycholemmal horn
145. Onycholysis = plummer's nails
146. Onychomadesis
147. Onychomatricoma
148. Pigmented onychomatricoma
149. Onychomycosis (distal and lateral subungual & superficial white & proximal subungual & mixed pattern & total dystrophic & endonyx)
150. Candida onychomycosis
151. Onychopapilloma
152. Onychophagia= Nail biting

153. Onychophosis =onychoclavus
154. Onychorehexis
155. Onychoschizia =lamellar splitting
156. Onychoteiromania
157. Onychotemnomania
158. Onychotillomania
159. Osteochondroma
160. Partial hereditary onycholysis
161. Acute Paronychia
162. Chronic Paronychia
163. Pachyonychia congenita
164. Parakertaotic/parakeratosis pustulosa (Hjorth-Sabouraud syndrome)
165. Parrot-beak nails
166. photo-onycholysis
167. Pincer nail=Trumpet nails
168. Pityriasis rubra pilaris :nail signs
169. Nail Pitting
170. Platonychia
171. Plicated nail
172. Poroma
173. Post-traumatic ectopic nail
174. Pseudomycotic nail dystrophy
175. Psoriasis : nail signs
176. Pterygium
177. Pyogenic granuloma
178. Racquet nails (nail en raquette)
179. Ragged cuticles
180. Red lunulae
181. Retronychia
182. Rheumatoid arthritis : nail signs
183. Rippled nail
184. Rosenau's depression

185.Round fingerpad
186. Rubronychia
187.Scabies: nail signs
188.Schwannoma
189.Scleromyxedema :nail signs
190.Seborrheic keratosis of nail
191.Shell nail syndrome
192.Splinter heamorhage
193.Nail Splitting
194.Squamous cell carcinoma (SCC)
195.Stippled nails
196.Subungual calcification
197.Subungual exostosis
198.Subungual heamatoma
199.Subungual heloma = corn
200.Subungual hyperkeratosis
201.Subungual melanoma
202.Subungual osteochondroma
203.Systemic lupus: nail signs
204.Systemic sclerosis: nail signs
205.Terry nail
206. Tile nail
207.Trachyonychia = Twenty nail syndrome
208.Transverse overcurvature of the nail
209.Trapezoidal nail
210.Triangular lunula
211.Tulip fingers
212.Upslanting toenail
213.Upward growth of nail
214.Wart
215.Worn down
216.Xanthogranuloma of nail
217.Xantholeukonychia

218.Xanthonychia
219.Yellow Lunula
220.Yellow nail syndrome

(2) Diseases of skin and mucous membranes

1. **Aarskog syndrome= Facial digital genital syndrome**
2. **ABCD syndrome (albinism, black lock, cell migration disorder of gut&deafness)**
3. **Abcess**
4. **Ablution dermatitis**
5. **Abrikossoff's tumour**
6. **Abuse (Child& Partner &Elder&Sexual)**
7. **Acatalasemia**
8. **Acanthamebiasis**
9. **Acanthoma/granuloma fissuratum**
10. **Acanthosis nigricans AN**
11. **Nevoid type Acanthosis nigricans**
12. **Acanthosis nigricans-like mycosis fungoides**
13. **Acantholytic acanthoma**
14. **Acantholytic dyskeratotic acanthoma ADA**
15. **Accessory digit= Supernumerary digit=Rudimentary Polydactyly**
16. **Accessory ear pinna**
17. **Accessory nipple= supernumerary nipples**
18. **Accessory oral cavity**
19. **Accessory tragus=preauricular tag=Supernumerary pinna**
20. **Achenbach syndrome=Paroxysmal hand hematoma**
21. **Achondroplasia =short limbed dwarfism**
22. **Aciduria**
23. **Acinic cell carcinoma**
24. **Acne aestivalis/ estival**
25. **Aquagenic acne**
26. **Acne chlorance**
27. **Acne conglobata**
28. **Acne cosmetica**
29. **Acne detergent**
30. **Acne excoriee=acne excoriée des jeunes filles**

31. **Acne-free naevus**
32. **Acne Fulminans**
33. **Acne Fulminans sine fulminans**
34. **Acne Halogens**
35. **Acne Hemorrhagica**
36. **Hormonal acne**
37. **Infantile acne**
38. **Acne keloidalis nuchea= folliculitis keloidalis nuchae= folliculitis scleroticans nuchae= Dermatitis papillaris capillitii**
39. **Maskne**
40. **Acne mallorca**
41. **Acne mcdonald**
42. **Acne mechanicha & fiddler's neck**
43. **Mid-childhood acne**
44. **Acne necrotica miliaris**
45. **Acne necrotica varioliformis=Necrotizing lymphocytic folliculitis**
46. **Neonatal Acne**
47. **Munro acne nevus**
48. **Oil acne**
49. **Acne pitch**
50. **Acne pomade**
51. **post Adolescent acne**
52. **Postmenopausal Acne**
53. **Preadolescent acne**
54. **Acne scar (ice pick&rolling&boxcar&pitted&stellate& crateriform scars& post acne erythema &keloid &lipoatrophic& papular&hypopigmented)**
55. **Senile Acne = Favre– Racouchot syndrome=solar comedones**
56. **Smoker Acne**
57. **Tar acne**
58. **Toilet acne**
59. **Tropical Acne**
60. **Acne rosacea**
61. **Acne due to radiation = Radiation-induced comedones**

62. **Acne scleroticans capitis**
63. **Acne vasculitis**
64. **Acne venenata**
65. **Acne vulgaris**
66. **Acneform drug eruption**
67. **Steroid induced acne**
68. **Acnitis =Acne agminata**
69. **Acquired bilateral melanosis of the neck in the perimenopausal woman (ABMNPW)**
70. **Acquired bilateral nevus of ota- like macules (ABNOM)**
71. **Acquired bilateral telangiectatic macules (ABTM)**
72. **Acquired blaschkoid dermatitis**
73. **Acquired Dermal Macular Hyperpigmentation**
74. **Acquired dermal melanocytosis**
75. **Acquired digital fibrokeratoma (ADFK)**
76. **Acquired dyskeratotic leukoplakia**
77. **Acquired elastotic hemangioma**
78. **Acquired facial blaschkoid melanocytic nevus**
79. **Acquired focal dermal elastosis**
80. **Acquired idiopathic generalized anhidrosis AIGA**
81. **Acquired immunodeficiency syndrome (AIDS)– HIV**
82. **Acquired linear hyperpigmentation of the lips (ALHL)**
83. **Acquired Palmoplantar keratoderma**
84. **Acquired perforating dermatosis =kyrle disease**
85. **Acquired progressive lymphangioma**
86. **Acquired progressive kinking of the hair=symmetrical circumscribed allotrichia**
87. **Acquired Raised Bands of Infancy**
88. **Acquired reactive digital fibroma**
89. **Acquired symmetrical acrokeratoderma**
90. **Acquired universal melanosis = carbon baby syndrome**
91. **Acral angioosteoma cutis**
92. **Acral arteriolar ectasia**

93. Acral fibrochondromyxoid tumor (AFCMT)
94. Acral fibromyxoma AFM
95. Superficial acral fibromyxoma (SAF)
96. Acral histiocytic nodules
97. Acral keratosis with eosinophilic dermal deposits
98. Acral myxoinflammatory fibroblastic sarcoma AMFS
99. Acral peeling skin syndrome
100. Acral pseudolymphomatous angiokeratoma APACHE
101. Acral psoriasiform hemispherical papulosis
102. Acral speckled hypomelanosis (ASH)
103. Acroangiodermatitis of Mali= pseudo-kaposi sarcoma
104. Acroangiodermatitis of Stewart bluefarb syndrome (SBS)
105. Acrocephalosyndactyly
106. Acrocyanosis
107. Acrodermatitis acidemica
108. Acrodermatitis chronica atrophicans of lyme
109. Acrodermatitis continua of Hallopeau=dermatitis repens
110. Acrodermatitis dysmetabolica
111. Acrodermatitis enteropathica
112. Acrodermatitis pustulosa
113. Acarodermatitis urticarioides
114. Acrodynia=epidemic erythema =calomel disease = pink disease=erythredema polyneuropathy
115. Acrodysostosis
116. Acrogeria=Gottron syndrome
117. Acrokeratoelastoidosis of Oswald Costa
118. Acrokeratosis paraneoplastica =Bazex syndrome
119. Acrokeratosis verruciformis of Hopf
120. Acromegaloid phenotype with cutis verticis gyrata and corneal leukoma
121. Acromelanosis
122. Acromelanosis albo-punctata
123. Achromia parasitica
124. Acroosteolysis

125. **Acroosteolysis with osteoporosis= Hajdu–Cheney syndrome**
126. **Acropathia ulcero-mutilans acquisita=Bureau–Barrière Thomas syndrome**
127. **Acropustulosis of infancy**
128. **Acrospiroma**
129. **Actinic comedonal plaque**
130. **Actinic granuloma o' brien**
131. **Actinic /senile/solar keratosis: (Atrophic & Actinic cheilitis & Hypertrophic (hyperkeratotic) & Lichenoid & Pigmented)**
132. **Chronic actinic dermatitis CAC = photosensitivity dermatitis= photosensitive eczema**
133. **Actinic reticuloid**
134. **Actinic prurigo= Hutchinson's summer prurigo =Familial hereditary polymorphous light eruption of american Indians**
135. **Acinetobacter-baumannii-associated Infection**
136. **Actinomycosis**
137. **Actinobacillosis**
138. **Activated pi3kδ syndrome**
139. **Acute and recurrent facial pustulosis**
140. **Acute bullous hemorrhagic prurigo**
141. **Acute cutaneous necrosis syndrome**
142. **Acute digital gangrene in newborn**
143. **Acute generalized exanthematous pustulosis AGEP = Toxic pustuloderma**
144. **Acute hemorrhagic edema of infancy AHEI= Finkelstein–Seidlmayer disease=Cockarde Purpura and Edema=Postinfectious cockade purpura**
145. **Acute inflammatory edema (AIE)=erythema of edema**
146. **Acute localized exanthematous pustulosis ALEP**
147. **Acute syndrome of apoptotic pan epidermolysis**
148. **Adam17 deletion=neonatal-onset inflammatory skin and bowel disease**
149. **Adams Oliver syndrome=Congenital scalp defects with distal limb anomalies**
150. **Adducted thumbs syndrome**
151. **Adeno virus related diseases**
152. **Adenocarcinoma**

153. **Adenoid cystic carcinoma**
154. **Adenolipoma**
155. **Adenomas in minor vestibular glands**
156. **Adenosine deaminase 2 (ADA) deficiency**
157. **Adenosquamous carcinoma**
158. **Adermatoglyphia**
159. **Adnexal adenocarcinoma not otherwise specified AANOS**
160. **Adnexal polyp of neonatal skin**
161. **Adrenoleukodystrophy= Schilder's disease**
162. **Adult gluteal granuloma=granuloma gluteale adultorum**
163. **Adult-onset still disease (AOSD)**
164. **AESOP syndrome : adenopathy, extensive skin patch overlying plasmacytoma**
165. **African tick bite fever**
166. **Aggressive digital papillary adenocarcinoma**
167. **Aggressive digital adenoma**
168. **Aggressive digital papillary adenoma**
169. **Agminated pigmented matricoma**
170. **Aicardi–goutières syndrome**
171. **Ainhum=Dactylolysis spontanea**
172. **Alagille syndrome**
173. **Albinism & oculocutaneous albinism (7types)**
174. **Albright hereditary osteodystrophy AHO**
175. **Aleukemic leukemia cutis**
176. **Alezzandrini syndrome**
177. **Algal infection**
178. **Alkaptonuria**
179. **Allgrove's syndrome**
180. **Alopecia-contractures-dwarfism mental retardation syndrome**
181. **Ant- induced alopecia**
182. **Accidental alopecia**
183. **Androgenetic alopecia AGA =Common baldness=Male pattern hair loss or Female pattern hair loss**

184. **Diffuse patterned alopecia (DPA)**
185. **Diffuse unpatterned alopecia (DUPA)**
186. **Reverse pattern androgenetic alopecia**
187. **Pseudo male pattern androgenetic alopecia**
188. **Alopecia areata =Area celsi (patchy&totalis &universalis &ophiasis & linear& perinevoid)**
189. **Alopecia areta ophiasis inversa= Sisaipho**
190. **Alopecia areata incognito**
191. **Central centrifugal cicatricial alopecia = Hot comb alopecia**
192. **Chemotherapy-induced alopecia CIA**
193. **Cicatricial /scarring alopecia**
194. **Cicatricial marginal alopecia CMA**
195. **Congenital Triangular Alopecia CTA= Brauer nevus**
196. **Alopecia of DLE discoid lupus erythematosus**
197. **Fibrosing alopecia in a pattern distribution (FAPD)**
198. **Frontal fibrosing alopecia FFA**
199. **Postmenopausal frontal fibrosing alopecia (PFFA)**
200. **Alopecia mucinosa**
201. **Alopecia neoplastica**
202. **Noncicatricial alopecia from systemic lupus erythematosus**
203. **Occipital Alopecia**
204. **Alopecia parvimaculata**
205. **Alopecia porphyrinica**
206. **Premature alopecia**
207. **pseudopelade of Brocq= Alopecia Atrophicans= Alopecia Cicatrisata**
208. **Pressure induced alopecia=post-operative alopecia**
209. **Psoriatic alopecia**
210. **Psoriasiform alopecia**
211. **Psychogenic alopecia**
212. **Radiation-induced alopecia**
213. **Retrograde alopecia**
214. **Senescent alopecia**
215. **Syphilitic alopecia SA**

216. **Toxic alopecia**
217. **Tractional alopecia**
218. **Traumatic alopecia**
219. **Congenital Alopecia universalis**
220. **Varicella-zoster virus (VZV) associated alopecia**
221. **Alopecic and aseptic nodule of scalp**
222. **Alopecia-onycho-dysplasia-hypohidrosis-deafness Syndrome**
223. **Alopecia–anosmia–deafness–hypogonadism (johnson neuroectodermal syndrome)**
224. **Anterolateral alopecia of the legs= peroneal alopecia**
225. **Alport syndrome**
226. **Alpha-gal syndrome**
227. **Alteranariosis**
228. **Alveolar soft part sarcoma**
229. **Amalgam tooth related diseases**
230. **Amalgam tattoo**
231. **Amaurosis congenita, cone–rod type, with congenital hypertrichosis**
232. **Amastia**
233. **Amazia**
234. **Amoebiasis cutis**
235. **Ameloblastoma**
236. **Amelo-onychohypohidrotic dysplasia**
237. **Amelo-cerebrohypohidrotic syndrome (kohlschutter–Tonz syndrome; epilepsy and yellow teeth syndrome)**
238. **American tick bite fever=Maculatum disease**
239. **Amicrobial pustulosis of fold**
240. **Amish brittle hair syndrome(BIDS):Brittle Hair&Intellectual Impairment&Decreased Fertility&Short Stature&Nail Dystrophy)**
241. **Amniotic constriction band (amnion rupture sequence)**
242. **Amputee-related dermatoses**

243. **Amyloidosis (see next)**
244. ✓ **Amyloidosis cutis dyschromia**
245. ✓ **Amyloid purpura**
246. ✓ **Anosacral**
247. ✓ **Atrophic nodular cutaneous Amyloidosis (ANCA)**
248. ✓ **Biphasic**
249. ✓ **Bullous**
250. ✓ **Dialysis related Amyloidosis**
251. ✓ **Familial Amyloidosis, finnish type**
252. ✓ **Familial cutaneous Amyloidosis**
253. ✓ **Familial Amyloidotic polyneuropathy**
254. ✓ **Frictional**
255. ✓ **Lichen /papular**
256. ✓ **Macular**
257. ✓ **Nodular**
258. ✓ **Poikiloderma-like cutaneous Amyloidosis**
259. ✓ **Primary Amyloidosis of the external ear (PAEE)=Auricular collagen papules**
260. ✓ **Systemic Amyloidosis (Primary &Secondary)**
261. **Amyloid elastosis**
262. **Anagen effluvium**
263. **Anasarca-induced desquamation**
264. **Ancylostomiasis**
265. **Anetoderma =Atrophia maculosa cutis (Primary: jadassohn–pellizzari & Schweninger-Buzzi anetoderma & secondary & of prematurity &familial)**
266. **Angelman syndrome**
267. **Angina bullous heamorhagica= Benign hemorrhagic bullous stomatitis**
268. **Angioedema**
269. **Angiofibroma**
270. **Angiohistiocytoma**
271. **Angioinvasive fungal infections**

272. **Angiokeratoma (Solitary &Circumscriptum & Mibelli & Corporis diffusum& Fordyce)**
273. **Angioleiomyoma**
274. **Angiolipoma**
275. **Angiolipoleiomyoma**
276. **Angiolymphoid hyperplasia with eosinophilia (ALHE)= Epithelioid hemangioma=pseudopyogenic granuloma=popular angioplasia =Histiocytoid hemangioma**
277. **Angioma serpignosa= progressive patchy capillary malformations**
278. **Angiomatosis**
279. **Angiomatoid fibrous histiocytoma=Angiomatoid malignant fibrous histiocytoma**
280. **Glomeruloid reactive angioendotheliomatosis**
281. **Malignant angioendotheliomatosis**
282. **Reactive angioendotheliomatosis (RAE)**
283. **Angiomyofibroblastoma**
284. **Angiomyolipoma**
285. **Angiomyomatous hamartoma**
286. **Superficial Angiomyxoma**
287. **Deep Angiomyxoma**
288. **Angiopericytomatosis**
289. **Angiosarcoma=Lymphangiosarcoma=Hemangiosarcoma**
290. **Angora hair nevus Syndrome =Schauder syndrome**
291. **Angry back syndrome**
292. **Anhidrosis**

Animals in dermatology (see next)

293. ✓**Alligator boy**
294. ✓**Bird-like facies**
295. ✓**Bovine popular stomatitis**
296. ✓**Buffalo hump**
297. ✓**Bulldog jaw**
298. ✓**Bull's eye appearance**

299. ✓ **Butterfly disease**
300. ✓ **Butterfly effect**
301. ✓ **Butterfly pox**
302. ✓ **Butterfly rash**
303. ✓ **Butterfly sign**
304. ✓ **Cat bite sign**
305. ✓ **Cat scratch disease**
306. ✓ **Cat tongue sign**
307. ✓ **Caterpillar bodies**
308. ✓ **Caterpillar dermatitis**
309. ✓ **Caviar appearance on dermoscopy of ochronosis**
310. ✓ **Caviar tongue**
311. ✓ **Chicken skin/Moroccan leather appearance**
312. ✓ **Chickenpox**
313. ✓ **Chicken wire appearance/pattern**
314. ✓ **Chicken wire erythema**
315. ✓ **Cowboy gunbelt and holster**
316. ✓ **Crab louse**
317. ✓ **Crab yaws**
318. ✓ **Crow's feet**
319. ✓ **Crowe's sign**
320. ✓ **Dalmatian dog'-like in fixed drug eruption**
321. ✓ **Dog-faced hypertrichosis lanuginose**
322. ✓ **Eggshell nails**
323. ✓ **Elephantiasis neurofibromatosa**
324. ✓ **Elephantine psoriasis**
325. ✓ **Elephantiasis nostras verrucosa**
326. ✓ **Elephantiasic variant thyroid dermopathy**
327. ✓ **faun tail nevus**
328. ✓ **Fish like scales**
329. ✓ **Fishman syndrome**

330. ✓Fish mouth wounds
331. ✓Fishnet-like pattern
332. ✓Fish odor syndrome
333. ✓Fish pedicure
334. ✓Fish skin graft
335. ✓Fish tail deformity
336. ✓Fish tank granuloma
337. ✓Fox–Fordyce disease
338. ✓Fried egg appearance
339. ✓Frog-spawn appearance
340. ✓Goose skin
341. ✓Horse rider's pernio
342. ✓Horseshoe-shaped' rim of hair
343. ✓Horse-riding stance
344. ✓Horse-collar pattern
345. ✓LAMB syndrome
346. ✓Leopard skin
347. ✓LEOPARD Syndrome
348. ✓Leonine facies
349. ✓Lizard skin
350. ✓Lobster claw deformity
351. ✓Mauskopf facies
352. ✓Millipede burns
353. ✓Moth-eaten alopecia
354. ✓Moth-eaten borders
355. ✓Moth-eaten appearance of the iris
356. ✓Moth-eaten appearance of skin
357. ✓Moth-eaten scales
358. ✓Monkey face
359. ✓Monkey facies
360. ✓Ostraceous psoriasis

361. ✓Owl's eye appearance
362. ✓Oyster-shell crusts
363. ✓Panda sign
364. ✓Parrot beak nail
365. ✓Parrot beak nose
366. ✓Pigskin appearance
367. ✓Plucked chicken appearance
368. ✓Pseudoparalysis of parrot
369. ✓Rabbit lines
370. ✓Rabbit burrow appearance
371. ✓Raccoon eyes/owl-eye sign
372. ✓Rat-tail macroconidia
373. ✓Rat-bite fever
374. ✓Rat mite itch
375. ✓Rodent ulcer
376. ✓ Rupioid psoriasis
377. ✓Russell's sign
378. ✓Salmon patch
379. ✓School of fish appearance
380. ✓Sea anemone appearance
381. ✓Shagreen patch
382. ✓Silver–Russell syndrome
383. ✓Snail track ulcers
384. ✓Spider angioma
385. ✓Starfish keratosis
386. ✓Stork bite
387. ✓Swan neck deformity
388. ✓Tadpole cells
389. ✓Tadpole seborrheic keratosis
390. ✓Toad skin
391. ✓Tiger tail pattern

392. ✓Turkey ear
393. ✓Turkey gobbler neck
394. ✓Ugly duckling sign
395. ✓Zebra bodies
396. ✓Zebra-like hyperpigmentation
397. ✓Zebra-stripe pattern
398. Ankyloglossia
399. Ankylostomiasis
400. Annular atrophic plaques of skin (Christianson's disease)
401. Annular elastolytic giant cell granuloma AEGCG =Actinic granuloma=Miescher granuloma of the face=O'Brien disease
402. Annular erythema of infancy
403. Annular lichenoid dermatitis of youth
404. Annually recurring erythema annulare centrifugum
405. Anogenital digitate keratosis
406. Anorexia nervosa with Diffuse reticulate purpura DRP
407. ANOTHER syndrome (alopecia, nail dystrophy, ophthalmic complications, thyroid dysfunction, hypohidrosis, ephelides and enteropathy, and respiratory tract infections)
408. Anthrax=Malignant pustule
409. Anti-3-hydroxy-3-methylglutaryl coenzyme A reductase immune-mediated necrotizing myopathy
410. Antiphospholipid syndrome
411. Anti-p105 pemphigoid
412. Anti-p200 Pemphigoid= Anti-laminin- γ [gamma]1 pemphigoid
413. Antithrombin iii deficiency
414. Antisynthetase syndrome
415. Apert syndrome= Acrocephalosyndactyly
416. Apical dysplasia of fingers
417. Aplasia cutis congenita =Congenital absence of skin : 9 types
418. Aphthous ulcer = Recurrent aphthous stomatitis (minor & major & herpetiformis)

419. **Apocrine carcinoma**
420. **Apocrine mixed tumors**
421. **Apocrine nevus**
422. **Aponeurotic fibroma**
423. **Aquagenic pruritus**
424. **Arborizing and stellate melanosis**
425. **Argininosuccinic aciduria**
426. **Argyria**
427. **Arsenicosis**
428. **Arsenic keratosis**
429. **Arsenical melanoderma**
430. **Arterial dissection plus lentiginosis**
431. **Arterial embolism**
432. **Arterial tortuosity syndrome**
433. **Arteriovenous fistula**
434. **Arteriovenous Haemangioma**
435. **Arteriovenous malformation**
436. **Arterio-veno-lymphatic malformations**
437. **Arthrogryposis**
438. **Arthrogryposis–Renal Dysfunction–cholestasis (ARC) syndrome**
439. **Ascariasis**
440. **Ascher syndrome**
441. **Aseptic abscess syndrome**
442. **Aspartylglucosaminuria**
443. **Aspergillosis**
444. **Astrakhan spotted fever**
445. **Ataxia telangiectasia= Louis-Bar syndrome**
446. **Athelia**
447. **Athlete's nodule**
448. **Athlete's foot**
449. **Atopic dermatitis /eczema**
450. **Atopic dirty neck**
451. **Atrichia with papular lesion**

452. **Atrophoderma elastolyticum discretum**
453. **Atrophoderma of Moulin**
454. **Atrophoderma of pasini and pierini =Sclérodermie atrophique d'emblée=Morphea plana**
455. **Atrophoderma vermiculata**
456. **Follicular atrophoderma**
457. **Atrophia maculosa varioliformis cutis**
458. **Atypical fibroxanthoma= Pseudosarcoma of the skin**
459. **Atypical lichenoid stomatitis**
460. **Atypical lipoma**
461. **Atypical Lipomatous Tumor**
462. **Atypical reaction to insect bite**
463. **Atypical vascular lesions**
464. **Auricular hematoma**
465. **Australian spotted fever**
466. **Autonomic denervation dermatitis**
467. **Autoimmune/inflammatory syndrome induced by adjuvants (ASIA syndrome)**
468. **Autoimmune lymphoproliferative syndrome (Canale–Smith syndrome)**
469. **Autoimmune polyendocrinopathy-candidiasis- ectodermal dystrophy) APECED**
470. **Autoimmune polyglandular syndrome : 4 Types**
471. **Axillary web syndrome**
472. **Babesiosis**
473. **Bacillary angiomatosis**
474. **Bafverstedt syndrome (benign circumscribed cutaneous lymphadenosis of the areola of the nipple)**
475. **Baisch syndrome**
476. **Baker's cyst**
477. **Balamuthia mandrillaris**
478. **Balanitis**
479. **Balanitis xerotica obliterans**
480. **Balanitis circumscripta plasmacellularis**

481. **Balanoposthitis**
482. **Balloon cell melanoma**
483. **Balloon cell nevus**
484. **BAP1 tumor predisposition syndrome (BAP1-TPDS)**
485. **Bap-oma**
486. **Bandler syndrome**
487. **Bannayan-Riley-Ruvalcaba syndrome**
488. **Bannayan-Zonana syndrome**
489. **Bannwarth syndrome**
490. **Barber say syndrome**
491. **Bart syndrome (aplasia cutis with epidermolysis bullosa)**
492. **Bartholin duct cyst**
493. **Basaloid follicular hamartoma BFH**
494. **Basal cell carcinoma BCC (see next) =Rodent ulcer**
495. ✓ **Nodular basal cell carcinoma**
496. ✓ **Pigmented type**
497. ✓ **Superficial basal cell carcinoma**
498. ✓ **Morpheaform basal cell carcinoma**
499. ✓ **Fibroepithelial basal cell carcinoma - Fibroepithelioma of pinkus**
500. ✓ **Micronodular basal cell carcinoma**
501. ✓ **Cystic basal cell carcinoma**
502. ✓ **Basosquamous carcinoma**
503. ✓ **Large Pore**
504. ✓ **Terebrant carcinoma**
505. ✓ **Polypoid**
506. ✓ **Basal cell nevus syndrome= Nevoid basal cell carcinoma syndrome =Gorlin -Goltz syndrome=Gorlin syndrome**
507. ✓ **Multiple hereditary infundibulo-cystic basal cell carcinoma syndrome**
508. **Basan syndrome**
509. **Basomelanocytic tumor**
510. **BASCULE syndrome (Bier anemic spots, cyanosis, and urticaria-like eruption)**

511. **Basidiobolomycosis**
512. **Bart-pumphrey syndrome**
513. **Bateman purpura**
514. **Bateman dermatitis**
515. **Battered child syndrome**
516. **Battered baby syndrome**
517. **Bazex- Dubre- Christol syndrome**
518. **Bazex syndrome**
519. **BCOR-CCNB3 undifferentiated sarcomas**
520. **Beals–Hecht syndrome= contractural arachnodactyly**
521. **Bean syndrome**
522. **Beare–Stevenson syndrome BSS**
523. **Betel leaf (piper betle)**
524. **Beckwith–wiedemann syndrome=Exomphalos-macroglossia -gigantism syndrome**
525. **Behcet disease**
526. **Bejel related disases=Endemic Syphilis**
527. **Bendar tumour**
528. **Benign alveolar ridge keratosis**
529. **Benign Follicular Hamartoma**
530. **Benign lymphoepithelial cyst**
531. **Benign mucinous metaplasia of the genitalia**
532. **Benign neonatal hemangiomatosis**
533. **Benign symmetric lipomatosis**
534. **Benign triton tumor =neuromuscular hamartoma =choristoma**
535. **Berardinelli's syndrome**
536. **Berlin syndrome**
537. **Berloque dermatitis= photo- toxic contact dermatitis**
538. **Berloque dermatitis(freckle like type)**
539. **Berk-Tabatznik syndrome**
540. **Besnier prurigo**
541. **Bicyclist's vulva**
542. **Bier spot =Angiospastic macules**

543. **Bilharziasis = Schistosomiasis**
544. **Biotin deficiency**
545. **Biotinidase deficiency**
546. **Bird nest hair**
547. **Bird-headed dwarfism= Seckel syndrome**
548. **Birt-Hogg-Dubé syndrome(alias Hornstein-Knickenberg syndrome)**
549. **Bites&stings&venoms(see next)**
550. **✓Arthropod bite**
551. **✓Anemon**
552. **✓Bee**
553. **✓Blister beetles/Rove beetle**
554. **✓lepidopterism**
555. **✓Bug**
556. **✓Cat**
557. **✓Caterpillar**
558. **✓Centipedes**
559. **✓Chigger**
560. **✓Ciguatera**
561. **✓Cockroaches**
562. **✓ Coral**
563. **✓Dog**
564. **✓Dogger Bank itch**
565. **✓Fire ant**
566. **✓Fish**
567. **✓Flea**
568. **✓Jelly fish**
569. **✓Leeches**
570. **✓Lizard**
571. **✓Locusta**
572. **✓Marine injuries**
573. **✓Mite**

574. ✓Mollusca (Octopuses and Conus Snails)
575. ✓Mosquito
576. ✓Portguese man of war =physalia physalis
577. ✓Sea bather ’s eruption
578. ✓Scorpion
579. ✓Snake
580. ✓ Spider
581. ✓Loxosceles Spiders
582. ✓Widow Spiders
583. ✓Tarantulas
584. ✓Tick
585. ✓Tongueworm
586. ✓Urchin
587. ✓Wasps
588. ✓Weever
589. Bizarre Parosteal Osteochondromatous Proliferation
590. Björnstad syndrome
591. Adult Blaschkitis
592. Acquired blaschkoid dermatitis
593. Blaschko linear acquired inflammatory skin eruption (BLAISE)
594. Blastomycosis=North american blastomycosis=Gilchrist’s Disease
595. Blau syndrome=juvenile systemic granulomatosis
596. Blepharochalsis
597. Blepharophyma
598. Blepharo-cheilo-dontic syndrome
599. Blistering dactylitis
600. Bloch-Sulzberger syndrome
601. Bloom syndrome=congenital telangiectatic erythema and stunted growth
602. Blue baby syndrome
603. Blueberry muffin baby
604. Blue man syndrome
605. Blue nevus

606. **Blue nevus-like metastatic melanoma**
607. **Blue rubber bleb nevus syndrome BRBNS= Bean syndrome**
608. **Blue toe syndrome**
609. **Blue tongue**
610. **Bockenheimer syndrome**
611. **Body dysmorphic disorder**
612. **Bohn 's nodule**
613. **Boil**
614. **Bonnet–Dechaume–Blanc syndrome**
615. **Book dysplasia**
616. **Book syndrome=Hyperhidrosis, premature canities and premolar aplasia**
617. **Börjeson–forssman–lehmann syndrome**
618. **Borrelial lymphocytoma cutis**
619. **Borrone dermato-cardio-skeletal syndrome**
620. **Botryomycosis=Bacterial pseudomycosis**
621. **Bowel-associated dermatosis–arthritis syndrome BADAS**
622. **Bourneville-pringle syndrome**
623. **Boutonneuse fever =Mediterranean Spotted Fever**
624. **Bowen disease=squamous cell carcinoma in situ**
625. **Pigmented Bowen disease**
626. **Bowenoid papulosis**
627. **Bowenoid actinic keratosis**
628. **BPES (blepharophimosis, ptosis, and epicanthus inversus syndrome**
629. **Brachioradial pruritus**
630. **Brachydactyly with absence of middle phalanges and hypoplastic nails or brachydactyly type A5 with nail dysplasia**
631. **Brachymetapody–anodontia–hypotrichosis–albinoidism(oculo-osteocutaneoussyndrome; Tuomaala syndrome**
632. **Brachymorphism-onychodysplasia-dysphalangism(BOD)**
633. **Branched-chain amino acid deficiency**
634. **Branchial cleft ,sinuses, and fistulae**
635. **Branchial Cleft cyst = Lymphoepithelial cyst= Lateral cervical cyst**
636. **Branchio-oculo-facial syndrome**

637. **Branchio-oto-renal syndrome**
638. **Brazilian hemorrhagic fever**
639. **BRESEK/BRESHECK syndrome(Brain anomalies-retardation of mentality and growth-ectodermal dysplasia-skeletal malformations-Hirschprung disease-ear deformity and deafness-eye hypoplasia-cleft palate-cryptorchidism-kidney dysplasia/hypoplasia**
640. **Bromhidrosis**
641. **Bromidrosiphobia**
642. **Bromoderma**
643. **Bronchogenic cyst**
644. **Bronze baby syndrome**
645. **Brooke-Spiegler syndrome**
646. **Brucellosis=Undulant fever**
647. **Brunsting-Perry pemphigoid (BPP)**
648. **Bruton syndrome**
649. **Bubble hair**
650. **Buccal exostoses**
651. **Buerger diseases**
652. **Buffalopox**
653. **Bulimia**
654. **postgraft Blisters**
655. **Bulla after burn**
656. **Coma bulla=Neurologic blisters**
657. **Bullous disease of diabetes=Bullosis diabeticorum**
658. **Edema Bulla= Hydrostatic bullae**
659. **Fracture bulla**
660. **Friction blister**
661. **Bullous hemorrhagic dermatosis**
662. **Bullous dermatitis artefacta**
663. **Bullous drug reactions**
664. **Bullous insect bite**
665. **Bullous neutrophilic dermatosis**
666. **Bullous urticaria**

667. **Bullous pemphigoid (see next)**
668. ✓ **Juvenile bullous pemphigoid**
669. ✓ **Brunsting–perry form**
670. ✓ **Dyshidrosiform pemphigoid**
671. ✓ **Eczematous pemphigoid**
672. ✓ **Erythrodermic pemphigoid**
673. ✓ **Large erosive ten-like lesions**
674. ✓ **Lichen planus pemphigoides**
675. ✓ **localized lesions**
676. ✓ **Papular pemphigoid**
677. ✓ **Pemphigoid nodularis**
678. ✓ **Pemphigoid vegetans**
679. **Bureau–Barrière syndrome**
680. **Burn**
681. **Cigarette burns**
682. **Burn Hemangioma (BH) =Scalded Pyogenic Granuloma**
683. **Aspirin ,Alcohol,Acrylic resin,Iodine : burn of oral cavity**
684. **Burning feet syndrome**
685. **Burning mouth syndrome**
686. **Burning scalp syndrome**
687. **Burning scrotum syndrome**
688. **Buschke-Lowenstein tumour**
689. **Buschke ollendorff syndrome**
690. **Bushy eyebrows**
691. **Byars-Jurkiewicz syndrome**
692. **Cactus granuloma**
693. **Cadasil syndrome (cerebral autosomal dominant Arteriopathy with subcortical infarcts and leuko-encephalopathy)**
694. **CAHMR syndrome (cataract, hypertrichosis, mental retardation)**
695. **Café-au-lait macules CALM**
696. **Cafe noir spots**
697. **Calabar swelling**

698. Calcinosis circumscripta
699. Calcinosis universalis
700. Tumoral calcinosis
701. Calcinosis cutis (Metastatic & Dystrophic & Idiopathic & Iatrogenic & Calcinosis universalis)
702. Perforating congenital calcinosis cutis of the ear
703. Calculus
704. Calciphylaxis= calcific uremicarteriolopathy=Calcifying Panniculitis
705. Calcifying aponeurotic fibroma
706. Calcifying fibrous pseudo-tumour
707. Caliber-persistent artery
708. Callosities
709. Callus
710. Camarena syndrome
711. Camelpox
712. Canalicular adenoma
713. Cancrum oris= Noma= Necrotising stomatitis
714. Candidiasis / Candidosis
715. ✓ Angular cheilitis (perlèche)=Angular stomatitis
716. ✓ Thrush (Acute pseudo-membranous candidiasis)
717. ✓ Chronic pseudomembranous candidiasis
718. ✓ Candidal leukoplakia/leukoplakia (chronic hyperplastic candidiasis)
719. ✓ Acute atrophic candidosis= Acute antibiotic stomatitis=Antibiotic sore mouth
720. ✓ Acute erythematous candidosis
721. ✓ Chronic denture stomatitis (chronic atrophic candidiasis)
722. ✓ Chronic erythematous candidosis
723. ✓ Chronic mucocutaneous candidiasis
724. ✓ Chronic nodular candidosis
725. ✓ Candida napkin/nappy dermatitis
726. ✓ Antibiotic (iatrogenic) candidiasis
727. ✓ Cheilocandidiasis

728. ✓Congenital cutaneous candidiasis
729. ✓Neonatal candidiasis
730. ✓Candidal vulvovaginitis
731. ✓Candidal Balanitis
732. ✓Candidal intertrigo=Flexural Candidiasis
733. ✓Candidal Miliaria
734. ✓Candidal Granuloma
735. ✓Candidid
736. ✓Candidemia
737. ✓Acute candidosis(Thrush& Acute antibiotic stomatitis)
738. ✓Chronic candidosis(Denture-induced stomatitis&Chronic hyperplastic candidosis&Chronic mucocutaneous candidosis&Erythematous candidosis)
739. CANDLE: chronic atypical neutrophilic dermatosis with lipodystrophy and elevated temperature
740. Canities
741. premature canities
742. Canities subita)
743. Cannon disease= white sponge naevus
744. Cantu syndrome= hypertrichotic osteochondrodysplasia
745. Capillary leak syndrome
746. Capillary malformation – arteriovenous malformation syndrome(CM-AVM syndrome)
747. Capnocytophaga canimorsus bacteremia
748. Carboxylase deficiency
749. Carbuncle
750. Carcinoid syndrome
751. Carcinosarcoma=Metaplastic carcinoma of the skin=Sarcomatoid Carcinoma
752. Carcinoma en cuirasse
753. Carcinoma erysipeloides
754. Carcinoma telangiectoides
755. Card14-mediated pustular psoriasis CAMPS

756. CARD-14 associated papulosquamous eruption (CAPE)
757. Cardio-acro-facial syndrome= Rabenhorst's syndrome
758. Cardiofaciocutaneous syndrome
759. Cardiomyopathy with alopecia and palmoplantar keratoderma (CAPK)
760. Carey syndrome
761. Carney complex
762. Carnitine translocase deficiency
763. Carotenemia
764. Carpenter syndrome
765. Cartilage hair hypoplasia= Mukusick-type metaphyseal chondrodysplasia
766. Cartilaginous papule of ear
767. Cartilagnous rest of neck
768. Carrion disease
769. Castleman diseases (giant lymph node hyperplasia, angiofollicular lymph node hyperplasia)
770. Cat scratch disease
771. Cataract–alopecia–sclerodactyly syndrome
772. Cattlepox
773. Cavernous hemangioma
774. Cavernous lymphangioma
775. CEDNIK: cerebral dysgenesis, neuropathy, ichthyosis, and keratoderma
776. Cellular digital fibroma
777. Cellulite= Gynoid lipodystrophy
778. Cellulitis=St Anthony's Fire
779. Helicobacter cellulitis
780. Cerebello -Trigeminal - Dermal dysplasia
781. Cerebro-Dermato-Osseous-Dysplasia
782. Cerebro-oculo-facio-skeletal (COFS)
783. Ceruminous adenoma
784. Ceruminous carcinoma
785. Cervical chondrocutaneous branchial remnants
786. Cysticercosis:Cestodes infections (Taenia saginata&Taenia solium)
787. Chagas disease= American trypanosomiasis

788. **Chancre of syphilis**
789. **Chancroid=soft chancres**
790. **CHANDS (curly hair, ankyloblepharon,nail dysplasia)syndrome**
791. **Charcot-Marie-Tooth disease**
792. **CHARGE syndrome = (coloboma, heart defects, atresia choanae, Growth retardation, genital abnormalities, and ear abnormalities)**
793. **Chediak - Higashi syndrome (CHS) =Béguez César–Steinbrinck syndrome**
794. **Angular cheilitis=Angular stomatitis=Perleche**
795. **Actinic cheilitis=Solar Cheilosis**
796. **Atopic cheilitis**
797. **Contact cheilitis**
798. **Drug induced cheilitis**
799. **Exfoliative cheilitis**
800. **Chronic loose scaly cheilitis**
801. **Factitial cheilitis**
802. **Granulomatous cheilitis=Miescher's cheilitis**
803. **Glandular cheilitis (see next)**
804. **✓Cheilitis Glandularis Apostematosa=Volkmann's cheilitis**
805. **✓superficial suppurative = Baelz-Unna**
806. **✓Glandular cheilitis simplex**
807. **Infective cheilitis**
808. **Nutritional cheilitis**
809. **Plasma cell cheilitis**
810. **Cheilitis simplex**
811. **Lip Chapping**
812. **Cheiralgia paresthetica**
813. **Cheiroarthropathy**
814. **Cherry angioma=Cherry hemangioma=Senile angioma=Campbell–demorgan spot**
815. **Cherubism**
816. **Cheyletiella dermatitis**
817. **Chickenpox =Varicella**

818. **Chickenpox scar**
819. **Neonatal varicella**
820. **Varicella gangrenosum**
821. **photolocalized Chickenpox**
822. **Chickenpox bullosa**
823. **Chikungunya- chicku**
824. **Childhood flexural comedones**
825. **Chlorotrichosis=Green hair**
826. **Chondrodermatitis nodularis helics**
827. **Chondrodermatitis nodularis nasi (of nose)**
828. **Chondromas= Soft Tissue Chondroma**
829. **Chondroblastic osteosarcoma**
830. **Chloroma=Granulocytic sarcoma**
831. **Chondroid lipoma**
832. **Chondroid syringoma= Mixed tumor of the skin**
833. **Malignant chondroid syringoma**
834. **Chondrosarcoma**
835. **chromhidrosis**
836. **Chromobacteriosis**
837. **Chromoblastomycosis=Chromomycosis=Fonseca's disease**
838. **Chronic bullous disease of childhood CBDC = linear IGA disease of childhood**
839. **Chronic eczematous eruptions of the aged**
840. **Chronic granulomatous disease**
841. **Chronic Idiopathic Penile edema**
842. **Chronic infantile neurological cutaneous and articular syndrome CINCa**
843. **Chronic recurrent annular neutrophilic dermatosis**
844. **Chronic nipple eczema**
845. **Chronic non-bacterial osteomyelitis**
846. **Chronic recurrent multifocal osteomyelitis**
847. **Chronic ulcerative stomatitis**
848. **Chrysiasis**

849. **Churg strausse vasculitis= Eosinophilic granulomatosis with polyangiitis=Allergic angiitis and granulomatosis**
850. **Cicatricial pemphigoid= Mucous Membrane Pemphigoid**
851. **Ciliated cyst of the vulva**
852. **Circle hairs**
853. **Circumscribed Palmar or plantar hypokeratosis**
854. **Circumferential skin creases Kunze type (CSC-KT)**
855. **Cisriod aneurysm**
856. **Citrullinemia**
857. **CLAPO syndrome (capillary malformation of the lower lip, lymphatic malformation predominant on the face and neck, asymmetry, and partial/generalized overgrowth)**
858. **Clarkson's syndrome**
859. **Clear cell acanthoma =acanthoma of Degos**
860. **Pigmented clear cell acanthoma**
861. **Clear cell hidradenoma (CCH)= Nodular hidradenoma**
862. **Clear cell hidradenocarcinoma**
863. **Clear cell papulosis**
864. **Clear cell squamous cell carcinoma CCSCC**
865. **Clear cell syringoma**
866. **Cleft lip**
867. **Cleft palate**
868. **Cleft palate–lateral synechia syndrome**
869. **Cleidocranial dysostosis with micrognathia, absent thumbs**
870. **Cleidocranial dysostosis with micrognathia, absent thumbs, and distal aphangia, Yunis–Varon syndrome**
871. **Cloacogenic carcinoma**
872. **CLOVES syndrome (Congenital, Lipomatous, Overgrowth, Vascular Malformations, Epidermal Nevi and Spinal/Skeletal Anomalies and/or Scoliosis)**
873. **Coagulopathies**
874. **Cobb syndrome=Cutaneomeningospinal angiomatosis**
875. **Cocaine-induced midline destructive lesion (CIMDL)**

876. **Coccidioidomycosis= San Joaquin valley fever= Posadas' Disease**
877. **Coccydynia**
878. **Cockayne syndrome**
879. **Coenurosis**
880. **Coffin–Siris syndrome**
881. **Coffin-Lowry syndrome**
882. **Cogan syndrome**
883. **Cohen syndrome**
884. **Coin-rubbing injuries**
885. **Cold agglutinin disease**
886. **Cold injury**
887. **Cole syndrome**
888. **Collagenous fibroma**
889. **Eruptive Collagenoma**
890. **Familial cutaneous Collagenoma**
891. **Isolated/solitary collagenoma**
892. **Collagenosis nuchae**
893. **Collagenous Papule of the Eyelid**
894. **Plantar cerebriform collagenoma**
895. **Papulolinear collagenoma=linear collagen nevus**
896. **Collagenous and elastotic plaques of the hands=degenerative collagenous plaques of the hands**
897. **Colloid milium= Elastosis colloidalis conglomerata=Miliary colloidoma**
898. **Collision tumour**
899. **Colorado tick fever**
900. **Comedocarcinoma**
901. **Comedo-like acantholytic dyskeratosis of the face and scalp**
902. **Open comedones**
903. **Microcomedone**
904. **Giant comedone**
905. **Macrocomedone**
906. **Submarine comedones**
907. **Sandpaper comedones**

908. **Double-edged comedones**
909. **Polyporous comedones**
910. **Compartment syndrome**
911. **Complement deficiency**
912. **Complex regional pain syndrome= Reflex sympathetic dystrophy**
913. **Composite hemangioendotheliomna**
914. **Compulsuive hand washing**
915. **Coenurosis**
916. **Condyloma acuminatum**
917. **Condyloma lata**
918. **Confetti-like macular atrophy**
919. **Confluent and reticulated papillomatosis Gougerot-Carteaud CARP**
920. **Congenital cd34+ granular cell dendrocytosis**
921. **Congenital comedones**
922. **Congenital deficiency of glutamine synthetase**
923. **Congenital diffuse melanosis**
924. **Congenital disorders of glycosylation**
925. **Congenital disseminated comedones**
926. **Congenital epulis / Epulides**
927. **Congenital erosive and vesicular dermatosis**
928. **Congenital fibrous hamartoma**
929. **Congenital hereditary endothelial dystrophy with nail hypoplasia**
930. **Congenital hypertrichotic melanoneurocytoma (CHM)**
931. **Congenital idiopathic generalized anhidrosis (CIGA)**
932. **Congenital infiltrating lipomatosis**
933. **Congenital ingrown toenails**
934. **Congenital insensitivity to pain CIP**
935. **Congenital insensitivity to pain with anhidrosis CIPA=HSAN type IV**
936. **Congenital linear atrophoderma**
937. **Congenital localized scleroderma**
938. **Congenital melanotic macule of the tongue (CMMT)**
939. **Congenital midline Cervical cleft**
940. **Congenital Perineal Groove**

941. **Congenital peristernal dermal sinuses**
942. **Congenital polypoid cutaneous adnexal and mesenchymal hamartoma CPCMH**
943. **Congenital pseudarthrosis of the clavicle**
944. **Congenital rubella syndrome**
945. **Congenital subungual pterygium**
946. **Congenital trichoid keratosis**
947. **Congenital vaccinia**
948. **Congenital varicella syndrome (CVS)**
949. **Congenital venous malformation**
950. **Conidobolus coronatus related Infections**
951. **Constitutional mismatch repair deficiency (CMMRD) syndrome**
952. **Contact stomatitis**
953. **Cooks syndrome**
954. **Copper (toxicity & deficiency) related signs**
955. **COPS (calcinosis cutis, osteoma cutis, poikiloderma, and skeletal abnormalities)**
956. **Corn**
957. **Cornelia de lange syndrome=typus amstelodamensis =Amsterdam dwarf=Brachmann-de Lange Syndrome**
958. **Corneodermato -osseous syndrome**
959. **Corona alopecia**
960. **Corona seborrhoeica**
961. **Corona veneris**
962. **Corona psoriatica**
963. **Costal fringe**
964. **Costello syndrome =facio-cutaneo-skeletal syndrome**
965. **Covid 19 (toes&fingers&COVID arm&COVID nails&COVID "red half‐moon nail" sign &COVID tongue&COVID mask&Kawa‐COVID‐19&COVID rash& COVID hand dermatitis)**
966. **Cowden syndrome**
967. **Cowpox**
968. **Coumadinn (coumarin, warfarin)-induced necrosis**

969. **Cranial fasciitis**
970. **Cranio ectodermal dysplasia=Sensenbrenner Syndrome**
971. **Craniofacial dysostosis with hypertrichosis and hypodontia**
972. **Craniofrontonasal dysplasia**
973. **Craniofaciocutaneous syndrome**
974. **Craniomandibular Dermatodysostosis =Familial Mandibulo-Acral Dysplasia**
975. **Craniosynostosis**
976. **Craniosynostosis ,anal anomalies and porokeratosis CAP syndrome**
977. **CREST syndrome (calcinosis, Raynaud phenomenon, esophageal dysmotility, sclerodactyly, and telangiectasia)**
978. **Cri-du-chat syndrome**
979. **Crimean congo hemorrhagic fever**
980. **Crohn's disease**
981. **Metastatic Crohn's disease**
982. **Cronkhite–Canada syndrome**
983. **Cross syndrome**
984. **Crouzon syndrome = Craniofacial dysostosis**
985. **Crow's feet**
986. **Crow-Fukase syndrome**
987. **CRTC1::TRIM11 cutaneous tumours (CTCT)**
988. **Cryoglobulinemia (3 types)**
989. **Cryofibrinogenemia**
990. **Crystalglobulin vasculopathy**
991. **Cryopyrin-associated periodic syndromes (CAPS)**
992. **Cryotherapy related complications**
993. **Cryptococcosis= European Blastomycosis**
994. **Cupping blisters**
995. **cutaneous arteriolitis**
996. **Cutaneous collagenous vasculopathy**
997. **Cutaneous ciliated cyst of lower limbs**
998. **Cutaneous endometriosis**
999. **Cutaneous eruption associated with streptococcus dysgalactiae bacteremia**

1000. **Cutaneous granulocytic sarcoma**
1001. **Cutaneous heterotopic bronchogenic tissue**
1002. **Cutaneous horn=Cornu cutaneum**
1003. **Cutaneous lymphadenoma**
1004. **Cutaneous M arundinis/P cyclothyrioides infections**
1005. **Cutaneous metallosis**
1006. **Cutaneous microsphaeropsis arundinis**
1007. **Cutaneous neosartorya hiratsukae infection**
1008. **Cutaneous paecilomyces infection**
1009. **Cutaneous T-cell dyscrasia (CTCD)**
1010. **Cutaneous waldenström macroglobulinemia**
1011. **Cutaneous yarrowia lipolytica infection**
1012. **Cutis laxa=Dermatochalasia= Dermatomegaly =Generalized elastolysis =Generalized elastorrhexis (see next)**
1013. **✓ADCL**
1014. **✓Progeroid ADCL (PADCL)**
1015. **✓ARCL type 1A**
1016. **✓ARCL type 1B**
1017. **✓ARCL type 1C(Urban–Rifkin–Davis syndrome)**
1018. **✓ARCL type 2A (Debré type; allelic to wrinkly skin syndrome)**
1019. **✓ARCL type 2B**
1020. **✓ARCL types 2C and 2D**
1021. **✓De Barsy syndrome A (ARCL type 3A)**
1022. **✓De Barsy syndrome B (ARCL type 3B)**
1023. **Cutis marmorata**
1024. **Cutis marmorata telangiectatica congenital=Van Lohuizen syndrome**
1025. **Cutis pleonasmus**
1026. **Cutis rhomboidalis nuchae**
1027. **Cutis tricolor**
1028. **Cutis verticis gyrate= pachydermia verticis gyrate**
1029. **Curry-Jones syndrome**
1030. **Cyanosis**

1031. **Cyclic edema**
1032. **Cyclical inguinal keratoderma**
1033. **Cydnidae =Burrowing bug**
1034. **Cylindroma**
1035. **Cylindrocarcinoma=malignant cylindroma**
1036. **Cyrano nose**
1037. **Cystadenoma**
1038. **Cystathionine synthetase deficiency**
1039. **Cysticercosis**
1040. **Cystic Condyloma**
1041. **Cystic fibrosis**
1042. **Cystic Neutrophilic Granulomatous Mastitis (CNGM)**
1043. **Cytomegalovirus infection (CMV)**
1044. **D-2-hydroxyglutaric aciduria**
1045. **Dabska's tumour**
1046. **Dacrocystitis**
1047. **Dadru**
1048. **Dandruff= pityriasis simplex capillitii**
1049. **Danthron erythema**
1050. **Darier white = Dyskeratosis follicularis =psorospermose folliculaire vegetante (see next)**
1051. **✓Guttate hypopigmentation**
1052. **✓Hyperpigmented flat-topped papules and macules=freckle-like**
1053. **✓Vesiculobullous lesions**
1054. **✓Acral hemorrhagic lesions**
1055. **✓Keratoderma**
1056. **✓ Cornifying**
1057. **✓Comedones**
1058. **✓Periocular nodulocystic lesions**
1059. **✓Alopecia**
1060. **✓cutis verticis gyrata**
1061. **Darier–Roussy disease**

1062. **Pseudo-darier's sign**
1063. **Dark halo/circles**
1064. **Darwin's tubercle**
1065. **Davener's dermatosis (DD)**
1066. **Deafness and ear pits**
1067. **Deafness, with vitiligo and muscle wasting**
1068. **De barsy syndrome**
1069. **Deep benign fibrous histiocytoma**
1070. **Deep dissecting hematoma (DDH)**
1071. **Deep venous thrombosis**
1072. **Defects of the interleukin-12/interferon-γ axis**
1073. **Defects in Toll-like receptor (TLR) signaling**
1074. **Deficiency of adenosine deaminase 2 (DADA2)**
1075. **Deficiency of interleukin-1 receptor antagonist DIRA**
1076. **Deficiency of interleukin-36 receptor antagonist DITRA**
1077. **Defoliants**
1078. **Degos disease = Malignant atrophic papulosis**
1079. **Delleman-Oorthuys syndrome**
1080. **Delusion parasitosis=Ekbom syndrome= Delusional infestation**
1081. **Delusional hyperhidrosis**
1082. **Demodicosis &demodex (see next)**
1083. ✓**Primary**
1084. ✓**Secondary**
1085. ✓ **pityriasis folliculorum=Facial Spinulate Demodicosis**
1086. ✓ **Periorificial dermatitis Demodicosis**
1087. ✓ **Facial dermatitis**
1088. ✓**Demodex folliculitis**
1089. ✓**Nodulocystic/conglobate demodicosis**
1090. ✓ **papulopustular demodicosis**
1091. ✓ **Demodex abscesses**
1092. ✓ **Ocular Demodicosis**
1093. ✓**Auricular Demodicosis**

1094. ✓ **Pigmented demodicosis**
1095. **Dengue fever**
1096. **Dento-oculocutaneous syndrome(Ackerman syndrome)**
1097. **Denture associated fibrous hyperplasia**
1098. **Denture stomatitis**
1099. **De sanctis–Cacchione syndrome**
1100. **Dental caries**
1101. **Dental sinus =Sinus of dental origin**
1102. **Dependency disease**
1103. **Dependent edema**
1104. **Dercum disease =Adiposis Dolorosa**
1105. **Dermal dendrocyte hamartomas**
1106. **Dermal dendrocyte hamartoma with stubby white hair**
1107. **Dermal duct tumour**
1108. **Dermal hyperneury**
1109. **Dermal melanocytic Hamartoma**
1110. **Dermal nerve sheath myxoma DNSM**
1111. **Dermatitis artefacta =factitial / factitious dermatitis =Pathomimia**
1112. **Dermatitis cruris pustulosa atrophican=folliculitis decalvans cruris=Sycosis Cruris**
1113. **Dermatitis herpetiformis DH= Duhring 's disease**
1114. **Dermatitis medicamentosa**
1115. **Dermatitis neglecta/ Dermatosis neglecta (DN)**
1116. **Dermatitis palmaris sicca**
1117. **Dermatitis passivata**
1118. **Dermatitis plantaris sicca**
1119. **Dermatitis repens**
1120. **Dermatitis simulata**
1121. **Dermatitis vegetans**
1122. **Dermatitis venenata**
1123. **Dermatodaxia**

1124. **Dermatofibroma DF =Benign fibrous histiocytoma= Fibroma simplex= Sclerosing haemangioma (see next)**
1125. **✓Aneurysmal dermatofibroma**
1126. **✓Ankle-type dermatofibroma (lipidized fibrous histiocytoma)**
1127. **✓Annular hemosiderotic dermatofibroma**
1128. **✓Delled ("atrophic") dermatofibroma**
1129. **✓Dermatofibroma with satellitosis**
1130. **✓Disseminated multiple dermatofibromas**
1131. **✓Erosive and ulcerated dermatofibroma**
1132. **✓Giant dermatofibroma**
1133. **✓Hyperkeratotic dermatofibroma**
1134. **✓Hyperpigmented nodular dermatofibroma**
1135. **✓Multinodular hemosiderotic dermatofibroma**
1136. **✓Multiple grouped dermatofibromas**
1137. **✓Nonpigmented dermatofibroma**
1138. **✓Pedunculated or polypoid dermatofibroma**
1139. **✓Subcutaneous dermatofibroma**
1140. **Dermatofibrosis lenticularis disseminate**
1141. **Dermatofibrohistiocytoma**
1142. **Dermatofibrosarcoma protuberans DFSP**
1143. **Pigmented dermatofibrosarcoma protuberans (Bednar)**
1144. **Congenital abnormalities of dermatoglyphs(Ridge aplasia, Ridge hypoplasia, Ridge dissociation, Ridges-off-the-end)**
1145. **Dermatographism (see next)=Dermatographia=Factitious Urticaria**
1146. **✓ Black dermatographism**
1147. **✓ Cholinergic dermatographism**
1148. **✓ Red dermatographism**
1149. **✓ White dermatographism**
1150. **✓ Yellow dermatographism**
1151. **✓Cold precipitated dermographism**
1152. **✓Delayed dermographism**

1153. ✓Follicular dermographism
1154. ✓Immediate dermographism
1155. Dermatoheliosis
1156. Dermatomycosis
1157. Dermatomyofibroma
1158. Dermatomyositis DM **(see next)**
1159. ✓ Adult-onset (Classic DM alone&Classic DM with malignancy&Classic DM as a part of an overlap connective tissue disorder&Clinically amyopathic DM (C-ADM)&Amyopathic DM (ADM)&Hypomyopathic DM)
1160. ✓ Juvenile onset (Classic DM&Clinically amyopathic DM (CADM) &Amyopathic DM (ADM)&Hypomyopathic DM)
1161. ✓Dermatomyositis sine myositis
1162. Dermatophilosis
1163. Dermatophytids
1164. Dermatopathia pigmentosa reticularis (DPR)
1165. Dermatophagia
1166. Dermatoporosis DP
1167. Dermatosis papulosis nigra DPN
1168. Dermatorynchus geneae
1169. Dermestid dermatitis =Carpet beetle dermatitis
1170. Dermacentor-borne necrosis erythema and lymphadenopathy (DEBONEL)
1171. Dermochondrocorneal syndrome= françois'syndrome
1172. Dermo-odontodysplasia
1173. Dermoid cyst
1174. Desmons syndrome
1175. Diabetic/ diabetes related disorders
1176. Diabetic dermopathy
1177. Diabetic Thick Skin =Cheiroarthropathy
1178. Didymosis aplasticosebacea
1179. Diffuse capillary malformation with overgrowth DCMO
1180. Diffuse dermal angiomatosis
1181. Diffuse lipomatosis

1182. **Digeorge's syndrome**
1183. **Digital Fibromatosis**
1184. **Digital papillary adenocarcinoma DPA**
1185. **Digital papular calcinosis**
1186. **Digital papular calcific elastosis**
1187. **Digital verrucous fibroangioma**
1188. **Digitalgia paresthetica**
1189. **Dilated pore of winer**
1190. **Dimple & skin fossae**
1191. **Diogenes syndrome**
1192. **Dipetalonemiasis**
1193. **Diphtheria**
1194. **Dirofilariasis**
1195. **Dissecting cellulitis= Folliculitis capitis abscedens et suffodiens=Hoffman's disease =Dissecting folliculitis**
1196. **Disseminated hypopigmented keratoses (DHK)**
1197. **Disseminated intravascular coagulation (DIC)**
1198. **Distichiasis**
1199. **Divry-Van Bogaert syndrome= Cortico-Meningeal Angiomatosis**
1200. **Dock 8 deficiency :dedicator of cytokinesis 8**
1201. **Donohue syndrome**
1202. **Donovanosis= Granuloma inguinale**
1203. **DOORS syndrome(deafness, onychodystrophy, osteodystrophy, and mental retardation)**
1204. **Dowling Degos disease (Classic &Follicular)**
1205. **Down syndrome**
1206. **Vesiculopustular eruption of transient myeloproliferative disorder in down syndrome**
1207. **Double eyebrow**
1208. **Dracunculiasis=Guinea worm disease= Dracontiasis**
1209. **Drug abuse**
1210. **Cutaneous adverse drug reactions**
1211. **Bullous drug reactions**

1212. **Drug exanthema - morbilliform drug eruption= maculopapular drug eruptions**
1213. **Drug reaction with eosinophilia and systemic symptoms DRESS**
1214. **Drug-associated bullous pemphigoid**
1215. **Drug-associated linear iga dermatosis**
1216. **Drug-induced Acanthosis nigricans**
1217. **Drug-induced Accelerated rheumatoid nodulosis**
1218. **Drug-induced Acne (DIA)**
1219. **Drug-induced Acneiform eruption (DIAE)**
1220. **Drug-induced Alopecia**
1221. **Drug-induced Erythroderma**
1222. **Drug-Induced Linear Iga Disease**
1223. **Drug-induced lupus erythematosus**
1224. **Drug-induced hyperpigmentation**
1225. **Drug-induced hypopigmentation**
1226. **Drug induced changes in hair color**
1227. **Drug-induced hirsutism**
1228. **Drug-induced hypertrichosis**
1229. **Drug-induced ichthyosis**
1230. **Interstitial granulomatous drug reaction (IGDR)**
1231. **Drug-induced nail abnormalities**
1232. **Drug-induced necrotizing purpura**
1233. **Drug-induced Palmoplantar keratoderma**
1234. **Drug-induced pemphigus**
1235. **Drug triggered pemphigus**
1236. **Drug-induced Photoallergic reaction**
1237. **Drug-induced phototoxic reaction**
1238. **Drug-induced pruritus**
1239. **Drug-induced pseudolymphoma= Lymphomatoid drug reaction**
1240. **Drug-Induced Pseudoporphyria**
1241. **Drug-induced sarcoidosis**
1242. **Drug induced scarring hair loss**
1243. **Drug-induced sweet's syndrome**

1244. **Drug-induced vasculitis**
1245. **Drug-induced vitiligo**
1246. **Anaphylactoid reaction= Kounis syndrome**
1247. **Fixed drug eruption FDE**
1248. **Injection site reaction**
1249. **Lichen planus pigmentosus (lpp) like eruption**
1250. **Lichenoid drug eruption**
1251. **Palifermin-associated papular eruption**
1252. **Pityriasiform drug eruption**
1253. **psoriasiform drug eruption**
1254. **Purpuric drug reactions**
1255. **Pustular drug reactions**
1256. **Scleroderma-like reactions to taxanes**
1257. **True serum sickness**
1258. **Serum sickness-like reaction**
1259. **Symmetrical drug-related intertriginous and flexural exanthema = Baboon syndrome**
1260. **Chemotherapy related cutanous manifestation**
1261. **✓Acral erythrodysesthesia**
1262. **✓Ara -c ear**
1263. **✓Burgdorf's reaction**
1264. **✓Chemotherapy- -induced oral mucositis**
1265. **✓Chemotherapy recall dermatitis**
1266. **✓Eccrine squamous syringometaplasia**
1267. **✓Epidermal dysmaturation**
1268. **✓Epidermal dystrophy**
1269. **✓Erythrodysesthesia**
1270. **✓Drug Extravasation**
1271. **✓Flexural erythematous eruption**
1272. **✓Hand foot skin reaction**
1273. **✓Hand foot skin syndrome**
1274. **✓Intertriginous eruption associated with chemotherapy**

1275. ✓Intertrigo-like eruption, chemotherapy induced
1276. ✓Palmar-plantar erythema
1277. ✓Palmar-plantar erythrodysesthesia
1278. ✓Toxic acral erythema
1279. ✓Toxic erythema of chemotherapy
1280. ✓Toxic erythema of the palms and soles

Drugs related cutanous manifestation:

1281. ✓Allopurinol hypersensitivity syndrome
1282. ✓Arsenic
1283. ✓BCR-ABL Tyrosine Kinase Inhibitors
1284. ✓Bleomycin
1285. ✓BRAF inhibitor
1286. ✓Clofazimine
1287. ✓Dapsone hypersensitivity syndrome
1288. ✓Epidermal growth factor inhibitors EGFRI
1289. ✓Hydroquin
1290. ✓Insulin
1291. ✓Mitogen-activated protein kinase (MAPK) inhibitors
1292. ✓MEK1 inhibitor
1293. ✓Mercury induced skin lesions
1294. ✓ Methotrexate-induced papular eruption
1295. ✓Methotrexate (MTX) toxicity
1296. ✓Minocycline hypersensitivity syndrome
1297. ✓Minoxidil
1298. ✓Proteasome inhibitors
1299. ✓Retinoid
1300. ✓Steroid
1301. Dubowitz syndrome
1302. Dupuytren contracture = Palmar Fibromatosis
1303. Dwarfism–alopecia–pseudoanodontia–cutis laxa
1304. Dwarfism, cerebral atrophy and keratosis pilaris

1305. **Acquired Brachial Cutaneous Dyschromatosis**
1306. **Dyschromatosis symmetrica hereditaria**
1307. **Dyschromatosis universalis hereditaria**
1308. **Dyssebacea**
1309. **Dyskeratosis congenita (Zinsser–Engman–Cole syndrome)**
1310. **Dysthesia**
1311. **Dysesthetic anogenital pain syndromes**
1312. **Ear pit =preauricular sinus**
1313. **Easy bruising syndrome=Inherited giant platelet disorders(Bernard–Soulier syndrome&Velocardial syndrome & Montreal platelet syndrome&May–Hegglin anomaly &Gray platelet syndrome&Sebastian syndrome&Epstein syndrome&Fechtner syndrome&Mediterranean macrothrombocytopenia)**
1314. **Ebola virus related diseases**
1315. **Eccrine angiokeratomatous hamartoma**
1316. **Eccrine angiomatous hamartoma EAH**
1317. **Eccrine nevus**
1318. **Eccrine squamous syringometaplasia**
1319. **Ecchymosis**
1320. **Echo virus related diseases**
1321. **Ecthyma simplex= Ulcerative impetigo**
1322. **Ecthyma contagiosum =Orf**
1323. **Ecthyma gangrenosum**
1324. **Ectodermal Dysplasia (see next)**
1325. **✓ ADULT: Acro-Dermato-Ungual-Lacrimal-Tooth**
1326. **✓ AEC :Ankyloblepharon, Ectodermal Dysplasia, Cleft lip/palate=Hay–wells syndrome)**
1327. **✓EEC ectodermal Dysplasia, ectrodactyly, cleft lip/palate**
1328. **✓Hidrotic ectodermal Dysplasia=clouston syndrome**
1329. **✓immunodeficiency, myopathy and ED**
1330. **✓ Pure hair and nail ectodermal dysplasia (PHNED)**
1331. **✓Ectodermal Dysplasia nectin 4**
1332. **✓Nectinopathies: cleft lip&palate**

1333. ✓ED, ectrodactyly and macular dystrophy (EEM)
1334. ✓ Rapp Hodgkin Syndrome RHS
1335. ✓ Acrorenal field defect, ED, and lipoatrophic diabetes (AREDYLD) syndrome
1336. ✓Hypohidrotic ED–immune deficiency
1337. ✓Hypohidrotic Ectodermal Dysplasia=Christ–Siemens–Touraine syndrome=X-linked hypohidrotic ectodermal Dysplasia (XLHED)
1338. ✓Autosomal Recessive Hypohidrotic Ectodermal Dysplasia
1339. ✓Autosomal Dominant Hypohidrotic Ectodermal Dysplasia
1340. ✓Hypohidrotic Ectodermal Dysplasia with hypothyroidism and agenesis of the corpus callosum
1341. ✓Hypohidrotic Ectodermal Dysplasia with hypothyroidism and ciliary dyskinesia (HEDH syndrome)
1342. ✓Ectodermal Dysplasia-skin fragility syndrome (ED-SFS) = McGrath syndrome
1343. ✓Ectodermal Dysplasia with palatal paralysis
1344. ✓Ectodermal Dysplasia with mental retardation and syndactyly
1345. ✓Ectodermal Dysplasia with syndactyly Syndrome (EDSS)
1346. ✓Ectodermal Dysplasia and neurosensory deafness (Mikaelian syndrome)
1347. ✓Ectodermal Dysplasia with adrenal cyst (odonto-onycho-hypohidrotic dysplasia with midline scalp defect)
1348. ✓Ectodermal Dysplasia with severe mental retardation
1349. ✓Ectodermal Dysplasia with natal teeth (Turnpenny type)
1350. ✓Ectodermal Dysplasia with distinctive facies and preaxial polydactyl
1351. ✓ED with onychogryphosis
1352. ✓Polycystic brain associated with Ectodermal Dysplasia
1353. ✓ED with short stature
1354. ✓lelis syndrome (hypohidrotic Ectodermal Dysplasia with acanthosis nigricans)
1355. ✓Margarita island Ectodermal Dysplasia
1356. ✓Arthrogryposis and ectodermal Dysplasia (tricho-oculo-dermo-vertebral

syndrome; Alves syndrome)

1357. ✓Mesomelic dwarfism –skeletal abnormalities –Ectodermal Dysplasia
1358. ✓Cranio-Ectodermal Dysplasia CED syndrome=Sensenbrenner syndrome
1359. ✓Ectodermal Dysplasia with Alopecia–onychodysplasia–hypohidrosis – deafness
1360. ✓Ectodermal Dysplasia with cataracts and hearing defects
1361. Ectomesenchymal chondromyxoid tumor
1362. Ectomesenchymoma
1363. Ectopic breast tissue
1364. Ectopic cilia
1365. Eczema= contact dermatitis
1366. Airborn contact dermatitis
1367. Allergic contact dermatitis= Allergic eczema (pigmented & lichenoid& erythema multiforme & pigmented purpura& pseudolymphomatous & granulomatous & pustular)
1368. Apron eczema
1369. Asteatotic Eczema = Eczema craquelé =Winter eczema =Winter itch=Desiccation dermatitis
1370. Baboon Eczema = systemic allergic contact dermatitis
1371. Car fitter dermatitis=Mechanic hand dermatitis
1372. Car Seat Dermatitis
1373. Circumostomy eczema
1374. Commode dermatitis= Toilet-seat dermatitis
1375. Discoid Eczema =Nummular dermatitis=Nummular eczema
1376. Oid-oid disease
1377. Disseminated Eczema= Autosensitization dermatitis
1378. Plant Dermatitis
1379. Post traumatic eczema
1380. Eczema due to war chemicals
1381. Dyshidrotic eczema =Pompholyx= Acute and recurrent vesicular hand dermatitis = Vesicular eczema
1382. Eczema: follicular type

1383. **Frictional lichenoid dermatitis=Dermatitis papulosa juvenilis=frictional lichenoid eruption=Sutton's summer prurigo=sandbox dermatitis =Dermatitis papulosa adultorum**
1384. **Hand eczema (see next)**
1385. **✓Pompholyx (vesicular eczema of palms and soles), dyshidrotic eczema**
1386. **✓Patchy vesiculosquamous eczema**
1387. **✓Hyperkeratotic palmar eczema (tylotic eczema)= eczema keratoticum**
1388. **✓Recurrent focal palmar peeling= desquamation en aires**
1389. **✓Discoid eczema (nummular eczema)**
1390. **✓Wear and tear dermatitis (housewives dermatitis)**
1391. **✓Ring eczema**
1392. **✓Finger tip eczema=Pulpitis**
1393. **✓Apron eczema**
1394. **✓Gut=slaughterhouse eczema**
1395. **✓Chronic acral dermatitis**
1396. **Irritant contact dermatitis (ICD) (see next)**
1397. **✓ Acute irritant contact dermatitis**
1398. **✓ Cumulative irritant contact dermatitis**
1399. **✓ Acute delayed irritant contact dermatitis**
1400. **✓ Irritant reaction irritant contact dermatitis**
1401. **✓ Asteatotic dermatitis**
1402. **✓ Traumatic irritant contact dermatitis**
1403. **✓ Pustular and acneiform irritant contact dermatitis**
1404. **✓ Non-erythematous irritant contact dermatitis**
1405. **✓ Subjective or sensory irritant contact dermatitis**
1406. **✓ Contact urticaria**
1407. **✓ Frictional irritant contact dermatitis**
1408. **✓Airborne Irritant Contact Dermatitis**
1409. **Infectious eczematoid dermatitis**
1410. **Lymphomatoid contact dermatitis**
1411. **Microbial eczema of the feet**

1412. **Eczema-like psoriasiform dermatitis**
1413. **Eczema Marginatum**
1414. **Eczema Herpeticum= kaposi varicelliform eruption= Herpetic eczema**
1415. **Eczema Coxsackium**
1416. **Eczema Molluscatum**
1417. **Eczema Monkeypoxicum**
1418. **Eczema Vaccinatum**
1419. **Palmoplantar eczema**
1420. **Slobber dermatitis**
1421. **Stasis dermatitis= gravitational dermatitis = varicose eczema =congestion eczema**
1422. **Systemic contact dermatitis**
1423. **Ehlers–Danlos syndrome =Cutis hyperelastica (see next)**
1424. **✓classic**
1425. **✓classic-like**
1426. **✓cardiac valvular**
1427. **✓Hypermobile**
1428. **✓Vascular**
1429. **✓kyphoscoliotic**
1430. **✓Musculocontractural**
1431. **✓Arthrochalasia**
1432. **✓Dermatosparaxis**
1433. **✓periodontal**
1434. **✓Brittle cornea syndrome**
1435. **✓Myopathic**
1436. **✓Spondylodysplastic**
1437. **✓Fibronectin -deficient eds**
1438. **✓ Tenascin -x deficient)**
1439. **✓Kresse syndrome (ehlers–danlos,progeroid type)**
1440. **✓Hernandez (ehlers–danlos-like) syndrome**
1441. **Elaioconiosis**

1442. **Elastoderma**
1443. **Elastofibroma dorsi**
1444. **Elastoma of Dubreuilh**
1445. **Elastoma=Elastic tissue nevus=Nevus elasticus**
1446. **Elastosis linearis rubra nasi**
1447. **Elastosis perforans serpiginosa**
1448. **Elastotic nodules of the ear**
1449. **Juvenile elastoma**
1450. **Papular elastorrhexis =Papular elastosis**
1451. **Perforating calcific elastosis**
1452. **Elejalde syndrome (neuroectodermal melanolysosomal disease)**
1453. **Elephantiasis nostras verrucosa**
1454. **Elephantiasic pretibial myxedema**
1455. **Elephantiasis neuromatosa**
1456. **Elephantine psoriasis**
1457. **Ellis–van creveld syndrome=chondroectodermal dysplasia**
1458. **Emberger syndrome**
1459. **Emboli**
1460. **Cholestrol Emboli**
1461. **Oxalate Emboli**
1462. **Emboli cutis medicamentosa= Nicolau's syndrome**
1463. **Emergomycosis**
1464. **Emmonsiosis**
1465. **Encephalocele**
1466. **Endocarditis related skin lesions**
1467. **Endocrine: Acromegaly related skin lesions**
1468. **Endocrine: Addison disease related skin lesions**
1469. **Endocrine: Cushing syndromen related skin lesions**
1470. **Endocrine: Hyperthyroidism related skin lesions**
1471. **Endocrine: Hypothyroidism related skin lesions**
1472. **Endocrine: Pheochromocytoma**
1473. **Endocrine mucin-producing sweat gland carcinoma (EMPSGC)**
1474. **Engraftment syndrome**

1475. **Enterobiasis (pinworms)**
1476. **Entomophtoromycosis= Subcutaneous phycomycosis (Basidiobolomycosis and Rhinoentomophtoromycosis)**
1477. **Envelopathies**
1478. **Eosinophilic annular erythema**
1479. **Eosinophilic dermatosis of hematologic malignancy (EDHM)**
1480. **Eosinophilic fasciitis**
1481. **Eosinophilic granuloma**
1482. **Eosinophilic mastitis**
1483. **Eosinophilic myalgia syndrome**
1484. **Eosinophilic panniculitis**
1485. **Eosinophilic pustular folliculitis**
1486. **Eosinophilic ulcer tongue= Eosinophilic nodule of the tongue**
1487. **Eosinophilic, polymorphic, and pruritic eruption associated with radiotherapy**
1488. **Epidermization of lip**
1489. **Epidermoid cyst= Infundibular cyst=Epidermal cyst=Epidermal inclusion cyst**
1490. **Verrucous epidermoid cysts**
1491. **Epidermodysplasia Verruciformis EDV**
1492. **Epidermolysis bullosa acquisita EBA=Acquired epidermolysis bullosa (see next)**
1493. **✓Non-inflammatory mechanobullous disease**
1494. **✓BP-like presentation**
1495. **✓Mucous membrane pemphigoid-like presentation**
1496. **✓Brunsting–Perry cicatricial pemphigoid-like presentation**
1497. **✓linear IgA bullous dermatosis-like presentation**
1498. **Epidermolysis bullosa simplex (EBS) (see next)**
1499. **✓EBS superficialis (ebss)**
1500. **✓Acral peeling skin syndrome**
1501. **✓Acantholytic superficialis (ebs acanth)**

1502. ✓**Skin fragility syndromes:**

- **Desmoplakin deficiency (ebs-desmoplakin; skin fragility–woolly hair syndrome)**
- **plakoglobin deficiency (ebs-plakoglobin;skin fragility–plakoglobin deficiency)**
- **plakophilin deficiency (ebs-plakophilin;skin fragility–ectodermal dysplasia syndrome)**

1503. ✓ **EBS due to klhl24 ad kelch-like protein 24**
1504. ✓**EBS, generalized severe (Dowling–Meara)= Epidermolysis bullosa herpetiformis**
1505. ✓ **EBS, generalized intermediate**
1506. ✓**EBS,other generalized ,koebner type**
1507. ✓ **EBS, localized (Weber–Cockayne)**
1508. ✓ **EBS with mottled pigmentation**
1509. ✓ **EBS with pyloric atresia ar plectin; α 6 β 4 integrin**
1510. ✓ **EBS, AR-BP230 deficiency ar bullous pemphigoid antigen 1 (bp230)**
1511. ✓ **EBS, AR-exophilin 5 deficiency**
1512. ✓ **EBS, AR-KRT14**
1513. ✓ **EBS, migratory circinate**
1514. ✓ **EBS-ogna ad plectin**
1515. ✓**EBS with muscular dystrophy(EBS-MD)**
1516. ✓**Lethal acantholytic epidermolysis bullosa**
1517. **Dystrophic Epidermolysis bullousa DEB (see next)**
1518. ✓ **RDEB variants: localized, pretibial, pruriginosa**
1519. ✓**RDEB, bullous dermolysis of the newborn**
1520. ✓**RDEB, generalized intermediate (non-Hallopeau-Siemens type)**
1521. ✓ **RDEB, generalized severe (Hallopeau–Siemens)**
1522. ✓**RDEB, inversa**
1523. ✓**RDEB centripetalis**
1524. ✓ **DDEB, bullous dermolysis of the newborn**
1525. ✓ **DDEB, generalized Intermediate DDEB**

1526. ✓ DDEB variants: acral, pretibial, pruriginosa, nails only
1527. ✓Bart syndrome
1528. ✓Albo-papuloid epidermolysis bullosa nigricans of Pasinin
1529. ✓Cockayne-Touraine syndrome
1530. **Junctional Epidermolysis bullousa JEB (see next)**
1531. ✓ JEB with pyloric atresia
1532. ✓carmi syndrome
1533. ✓JEB with respiratory and renal involvement ar α 3 integrin subunit
1534. ✓JEB miti, generalized intermediate =Non herlitz generalized =Generalized atrophic benign epidermolysis bullosa GABEB= Epidermolysis bullosa atrophicans generalisata mitis
1535. ✓JEB, generalized severe Gravis (letalis,Herlitz disease)
1536. ✓ JEB, inversa
1537. ✓ JEB, late onset
1538. ✓JEB, localized
1539. ✓ JEB-laryngo-onycho-cutaneous syndrome=Shabbir's syndrome
1540. Epidermolytic acanthoma
1541. Cutaneous epithelioid angiomatous nodule (CEAN)
1542. Epithelioid hemangioma
1543. Epithelioid hemangioendothelioma
1544. Epithelioid cell histiocytoma= Epithelioid fibrous histiocytoma
1545. Epithelioid sarcoma
1546. Epithelioid sarcoma-like hemangioendothelioma (pseudomyogenic hemangioendothelioma)
1547. Epithelioma cuniculatum
1548. Episodic angioedema with eosinophilia=Gleich syndrome

Eponymous signs in dermatology (see next)

1549. ✓Albright's dimple sign
1550. ✓Alcohol wipe sign
1551. ✓Anal groove sign
1552. ✓Antenna sign

1553. ✓Apple jelly sign
1554. ✓Asboe-hansen sign
1555. ✓Auspitz sign
1556. ✓Axillary ecchymosis sign
1557. ✓Ball sign
1558. ✓Ball-SITE sign
1559. ✓Bare underbelly sign
1560. ✓Barnett's sign
1561. ✓Battle's sign (mastoid ecchymosis)
1562. ✓Beauty and the beast sign
1563. ✓Benediction sign
1564. ✓Berliner sign
1565. ✓Besnier's sign
1566. ✓Biederman's sign
1567. ✓ Biett's sign
1568. ✓ Bolognia sign
1569. ✓ Breakfast, lunch, and dinner sign
1570. ✓Blink sign
1571. ✓Blue dot sign
1572. ✓Blue pseudoveil sign
1573. ✓Blue underpants' sign
1574. ✓Blueberry muffin sign
1575. ✓Bonbon toffee sign
1576. ✓Borsieri's sign
1577. ✓Bote sign
1578. ✓Branham's sign (nicoladoni sign)
1579. ✓Bravermans sign
1580. ✓Brenner sign
1581. ✓Brownie nose sign
1582. ✓Bull's head sign
1583. ✓Burnt matchstick sign

1584. ✓**Buschke-ollendorff sign**
1585. ✓**Butterfly sign**
1586. ✓**Buttonhole sign**
1587. ✓**Calabar sign**
1588. ✓**Candle wax sign**
1589. ✓**Caput medusae**
1590. ✓**Carpet tack sign (tin tack sign, cat tongue sign)**
1591. ✓**Casal's necklace sign**
1592. ✓**Cayenne pepper sign**
1593. ✓**Central dell sign**
1594. ✓**Chagas–mazza–romaña's sign**
1595. ✓**Chandelier's sign**
1596. ✓**Chik sign**
1597. ✓**Clown nose sign**
1598. ✓**Cluster of jewels sign (string of pearls, rosette sign**
1599. ✓**Color-transition sign**
1600. ✓**Comby sign**
1601. ✓**Comet sign**
1602. ✓**Coral bead sign**
1603. ✓**Corn flake sign**
1604. ✓**Coudability sign**
1605. ✓**Coup d'ongle sign**
1606. ✓**Crowe's sign**
1607. ✓**Cuff sign**
1608. ✓**Cullen's sign**
1609. ✓**Cumulus sign**
1610. ✓**Darier's sign**
1611. ✓**Deck-chair sign**
1612. ✓**Delmege sign**
1613. ✓**Delta wing jet with contrail sign**
1614. ✓**Dew drop on rose petal sign**

1615. ✓Diamond necklace sign
1616. ✓Dimple sign (fitzpatrick sign)
1617. ✓Dirty neck sign
1618. ✓Dot-in-circle sign
1619. ✓Double edge sign
1620. ✓Drip sign
1621. ✓Dubois sign
1622. ✓Ear lobe sign
1623. ✓Ear sign
1624. ✓Eastern pancake sign
1625. ✓Elbow sign
1626. ✓Enamel paint sign (crazy pavement sign= crackled sign= mosaic sign= flaky paint sign)
1627. ✓Eyelid sign
1628. ✓Fibula sign
1629. ✓Filipovitch's sign
1630. ✓Flag sign – clinical
1631. ✓Flag sign-histopathological
1632. ✓Flambeau sign
1633. ✓Forscheimer's sign
1634. ✓Fountain sign
1635. ✓Fountain of youth sign
1636. ✓Frank's sign=Diagonal earlobe crease
1637. ✓Friar tuck sign
1638. ✓Fried egg sign
1639. ✓Frog spawn appearance
1640. ✓Froment sign
1641. ✓Frosted Glass Sign
1642. ✓Giovannini sign
1643. ✓Goat eye sign
1644. ✓Goldstein's toe sign

1645. ✓Golf club set sign
1646. ✓Gorlin's sign
1647. ✓Gottron's sign
1648. ✓Greenhow's sign (vagabond's sign)
1649. ✓Grey turner's sign
1650. ✓Groove sign of greenblatt
1651. ✓Groove sign
1652. ✓Gulliver's sign
1653. ✓Hair collar sign
1654. ✓Hairy palm sign
1655. ✓Halo sign
1656. ✓Hamburger sign
1657. ✓Handle bar sign
1658. ✓Hang-glider sign
1659. ✓Hanging groin sign
1660. ✓Hanging curtain sign
1661. ✓Hatchcock sign
1662. ✓Headlight sign
1663. ✓Heliotrope sign
1664. ✓Hertoghe's sign (queen anne's sign)
1665. ✓Hiker's feet sign
1666. ✓Hildreth's sign
1667. ✓Hoagland sign
1668. ✓Holster sign of dermatomyositis
1669. ✓Hutchinson's sign (melanoma &herpes zoster)
1670. ✓Hypopyon sign
1671. ✓Iceberg sign
1672. ✓Ice-pack sign
1673. ✓Igneous rock sign
1674. ✓Ingram's sign
1675. ✓Isobar sign

1676. ✓Jacquet's sign
1677. ✓Jellinek's sign (rasin's sign)
1678. ✓Jelly sign
1679. ✓Kaposi-stemmer sign
1680. ✓Keining sign
1681. ✓Kerr's sign
1682. ✓Knife cut sign
1683. ✓Lafora's sign
1684. ✓Leopard skin sign
1685. ✓Leser–trelat sign
1686. ✓Liddle's sign
1687. ✓Lightening sign
1688. ✓Linear gingival erythema sign
1689. ✓Little red riding hood sign
1690. ✓Lizard skin sign
1691. ✓Love's sign
1692. ✓Lyon's sign
1693. ✓Mace sign
1694. ✓Maroni's sign
1695. ✓Matchbox sign
1696. ✓Meffert's sign
1697. ✓Mephisto sign
1698. ✓Micro-hutchinson sign
1699. ✓Milian's ear sign
1700. ✓Milk sign (scarlet milk sign)
1701. ✓Mistletoe sign
1702. ✓Mizutani's sign (round finger pad sign)
1703. ✓Muehrcke's sign
1704. ✓Mushroom cloud sign
1705. ✓Myer sign
1706. ✓Nagayama sign

1707. ✓ **Nail flag sign**
1708. ✓ **Nasal crease sign**
1709. ✓ **Nazzaro's sign**
1710. ✓ **Neighborhood sign**
1711. ✓ **Nikolskiy's sign**
1712. ✓ **Noodle sign**
1713. ✓ **Nose sign (pavithran's nose sign)**
1714. ✓ **Oil drop sign**
1715. ✓ **Oil spot sign**
1716. ✓ **Osler's sign**
1717. ✓ **Ovoid palatal sign**
1718. ✓ **Oyster blister sign**
1719. ✓ **Paired ear creases of the helix sign**
1720. ✓ **Panda and lambda sign**
1721. ✓ **Pastia's sign**
1722. ✓ **Pathergy sign**
1723. ✓ **Patrick yesudian sign**
1724. ✓ **Pearl necklace sign**
1725. ✓ **Pemberton's sign**
1726. ✓ **Peripilar sign**
1727. ✓ **Pink glow sign**
1728. ✓ **Pink rim sign**
1729. ✓ **Pink sign**
1730. ✓ **Pitaluga's sign**
1731. ✓ **Pluck out sign**
1732. ✓ **Poppyfield bleeding sign**
1733. ✓ **Pore sign**
1734. ✓ **Prayer sign**
1735. ✓ **Premlatha's sign**
1736. ✓ **Promontory sign**
1737. ✓ **Punshi's sign**

1738. ✓**Pup-tent sign**
1739. ✓**Raccoon sign**
1740. ✓**Radial crown sign**
1741. ✓**Rainbow sign**
1742. ✓**Red planet sign**
1743. ✓**Red, white, and blue sign**
1744. ✓**Regularly bended ribbon sign**
1745. ✓**Reverse namaskar sign**
1746. ✓**Ring scales sign**
1747. ✓**Romana's sign**
1748. ✓**Rope sign**
1749. ✓**Rose spots**
1750. ✓**Rosette sign**
1751. ✓**Rudolph sign**
1752. ✓**Russell's sign**
1753. ✓**Sagrada familia sign, digitations and mirror sign**
1754. ✓**Salute sign**
1755. ✓**Samitz's sign**
1756. ✓**Sandwich sign**
1757. ✓**School chair sign**
1758. ✓**Scratch sign (coup d'ongle sign, besnier's sign)**
1759. ✓**Seat rash sign**
1760. ✓**Setting sun sign**
1761. ✓**Shar‐pei sign**
1762. ✓**Shawl sign**
1763. ✓**Shelly's sign**
1764. ✓**Shoulder pad sign**
1765. ✓**Shuster's sign**
1766. ✓**Skin crease sign**
1767. ✓**Snakeskin sign**
1768. ✓**Spade sign**

1769. ✓St.tropez sign
1770. ✓Stabler's and Bryant's sign
1771. ✓Stafne's sign
1772. ✓Starburst sign
1773. ✓Starry night sky sign
1774. ✓Statue of liberty sign
1775. ✓Sternberg's thumb sign
1776. ✓Stinson sign
1777. ✓String of pearls sign
1778. ✓Tap sign
1779. ✓Tasleem's water jet sign
1780. ✓Tent sign
1781. ✓Thompson's sign
1782. ✓Thumb sign (steinberg sign)
1783. ✓Thumbprint sign
1784. ✓Toothbrush sign
1785. ✓Toy soldier sign
1786. ✓Translucency sign
1787. ✓Triangular nasal notch sign
1788. ✓Triangular sign
1789. ✓Tripe palms sign
1790. ✓Ugly duckling sign
1791. ✓Umbilical sign
1792. ✓Urschel's sign
1793. ✓V- sign
1794. ✓Vieira sign
1795. ✓Volcano sign
1796. ✓Wake sign
1797. ✓Walker- Murdoch's/Wrist sign
1798. ✓Walzel's sign
1799. ✓Wartenberg's sign

1800. ✓White islands in a sea of red sign
1801. ✓Wimberger's sign
1802. ✓Winterbottom's sign
1803. ✓Wipeout sign
1804. ✓Wobble sign
1805. ✓Wrist sign (walker's sign)
1806. ✓Yellow clod sign
1807. ✓Yoga sign

Clothing-related Eponyms (see next)

1808. ✓Cradle cap
1809. ✓Turban tumors
1810. ✓Woolly hair
1811. ✓Spun glass hair
1812. ✓Beaded/necklace hair
1813. ✓Hidden hair
1814. ✓Hair collar sign
1815. ✓Hair pin vessels
1816. ✓Crown vessels
1817. ✓Lacy striae
1818. ✓Velvety plaques
1819. ✓Moroccan leather
1820. ✓Silk road disease=behcet disease
1821. ✓Harlequin armor
1822. ✓Bathing suit appearance
1823. ✓Bare underbelly sign
1824. ✓Shoulder pad sign
1825. ✓Shawl sign
1826. ✓Mask of pregnancy
1827. ✓Blue grey veil
1828. ✓Buttonhole sign
1829. ✓Gaiter region

1830. ✓Holster sign
1831. ✓Mitten hands/feet
1832. ✓Casal's necklace
1833. ✓Necklace of venus
1834. ✓Epstein pearls
1835. ✓String of pearls sign
1836. ✓Pearly appearance
1837. ✓Amniotic band syndrome
1838. ✓Keratodermic sandals
1839. ✓Beaded sign
1840. ✓Coral beads sign
1841. ✓Signet ring sign
1842. ✓Brass knuckles sign
1843. ✓Safety pin sign
1844. ✓Chaining pattern
1845. ✓Perivascular cuffing
1846. ✓Cuff sign
1847. ✓Rosette sign
1848. ✓Papular-purpuric gloves and socks syndrome

Food Eponyms in Dermatology(see next)

1849. ✓Apple jelly nodules
1850. ✓Bean bag cells
1851. ✓Bean' syndrome
1852. ✓Black berry stomatitis
1853. ✓Blueberry muffin baby
1854. ✓Blueberry muffin lesions
1855. ✓Breakfast, lunch and dinner sign
1856. ✓Café-au-lait macules
1857. ✓Cayenne pepper spots
1858. ✓Champagne bottle leg
1859. ✓Cherry angiomas

1860. ✓Corn flake sign
1861. ✓Curry-Hall syndrome
1862. ✓Curry Jones syndrome
1863. ✓Doughnut (sign& wart& granuloma)
1864. ✓Fried egg
1865. ✓Garlic clove fibroma
1866. ✓GEMSS syndrome
1867. ✓Grain itch/Barley itch/straw itch/Mattress itch/Prairie itch/cotton weed dermatitis
1868. ✓Honey comb atrophy
1869. ✓Mango dermatitis
1870. ✓Milian citrine skin
1871. ✓Mulberry like erosions
1872. ✓Mulberry molars
1873. ✓Mushroom deformity
1874. ✓Oil acne
1875. ✓Oil granuloma
1876. ✓Oil spot
1877. ✓Onion skin cysts
1878. ✓Peau d' orange
1879. ✓Port wine stain
1880. ✓Red Strawberry tongue
1881. ✓Salt and pepper fundus
1882. ✓Sandwich sign
1883. ✓Sausage finger
1884. ✓Spaghetti and meat balls
1885. ✓Strawberry gums
1886. ✓Strawberry hemangioma
1887. ✓Sweet syndrome
1888. ✓Swiss cheese effect
1889. ✓Toasted skin syndrome

1890. ✓Tomato catsup fundus
1891. ✓Wafer like scales
1892. ✓Watermelon stomach
1893. ✓White strawberry tongue

Eponyms of substance abuse (see next)

1894. ✓Cannabis arteritis
1895. ✓Betel chewer's mucosa
1896. ✓Chingari sign
1897. ✓Harlequin nails=quitters' nails
1898. ✓Smokers' palate=smokers' keratosis
1899. ✓ Smoker's tongue
1900. ✓Smokers' melanosis
1901. ✓Smoker's face
1902. ✓Smokers' moustache
1903. ✓Smokers' comedones
1904. ✓Smokers callosity
1905. ✓Snuff dippers lesions
1906. ✓Cocaine bugs
1907. ✓Cocaine-induced midline destructive lesions (cimdl)
1908. ✓Cocaine keratosis
1909. ✓Parrot beak nails
1910. ✓Formication
1911. ✓Gummies
1912. ✓Pseudovasculitis
1913. ✓Levamisole purpura
1914. ✓Matchbox sign
1915. ✓Snorters' warts
1916. ✓Ecstasy pimples
1917. ✓Meth mouth
1918. ✓Pentazocine ulcers
1919. ✓Pentazocine ulcers

1920. ✓High pruritus
1921. ✓Pseudoacanthosis nigricans
1922. ✓Puffy hands syndrome
1923. ✓Railroad tracks
1924. ✓Skin popping
1925. ✓Shooter's patch
1926. ✓Shooting tattoo
1927. ✓Track mark
1928. ✓Torniquet hyperpigmentation
1929. ✓Coma blisters
1930. ✓Glue sniffers rah

Syphilis signs(see next)

1931. ✓ Buschke-ollendorff sign
1932. ✓ Barber-pole sign
1933. ✓ Biederman's sign
1934. ✓Bucket handle sign
1935. ✓ Bulldog jaw sign
1936. ✓ Café-au-lait tint sign
1937. ✓ Crown of venus sign
1938. ✓Dermarquay sign
1939. ✓ Dory flop sign
1940. ✓ Du bois sign
1941. ✓ Elliot sign
1942. ✓ Higoumenaki's sign
1943. ✓ Hutchinson teeth sign
1944. ✓ Krisowski's sign
1945. ✓ Mulberry molars
1946. ✓ Natiform skull sign
1947. ✓ Olympian brow sign
1948. ✓ Omnibus sign
1949. ✓ Opera-glass nose sign

1950. ✓ Parrot's nodes
1951. ✓ Sabre shins (fournier's sign)
1952. ✓Sarcophagus sign
1953. ✓ Seeping sign
1954. ✓ Silex's sign
1955. ✓ Split papules
1956. ✓ Virchow's sign
1957. Epstein pearl
1958. Epstein barr virus - infectious mononucleosis =Glandular Fever= Pfeiffer Glandular Fever= Filatov's Disease
1959. Epstein-Barr Virus-Associated Smooth Muscle Tumors
1960. Epulis fissuratum / Epulides
1961. Epulis granulomatosa / Epulides
1962. Ethylmalonic aciduria
1963. Erosive adenomatosis of the nipple
1964. Erosive papulonodular dermatosis
1965. Erosive pustular dermatosis of the scalp
1966. EGFRI induced erosive pustular dermatosis of the scalp
1967. Erosive pustular dermatosis of the leg
1968. Eruptive angiomas associated with graft-versus-host disease
1969. Eruptive hypomelanosis (idiopathic)
1970. Eruptive keratoderma of the hands and feet of Rahim
1971. Eruptive lingual papillitis (ELP)
1972. Eruptive pseudoangiomatosis
1973. Eruptive recurrent pigmented telangiectasia
1974. Erysipelas
1975. Erysipeloid= Erysipeloid of Rosenbach
1976. Erythema
1977. Erythema ab igne = Toasted skin syndrome = Fire stains
1978. Erythema annulare centrifugum (EAC):superficial &deep variants
1979. Erythema arthriticum epidemicum
1980. Erythema chronicum migrans of lyme disease

1981. **Erythema contusiforme**
1982. **Erythema Dyschromicum perstans =Ashy dermatosis**
1983. **Erythema elevatum diutinum EED**
1984. **Erythema gyratum atrophican of neonate**
1985. **Erythema gyratum repen**
1986. **Erythema gyratum repen like urticarial vasculitis**
1987. **Erythema gyratum repen like bullous lupus**
1988. **Erythema gyratum repen like Erythema annulare centrifugum**
1989. **Erythema gyratum repen like psoriasis**
1990. **Erythema induratum (nodular vasculitis =erythema induratum of Bazin or Bazin disease (tuberculous etiology) & erythema induratum of whitfield (non-tuberculous etiology)**
1991. **Erythema infectiosum= fifth disease=slapped cheek syndrome**
1992. **Erythema marginatum**
1993. **Erythema marginatum-like lesions of hereditary angioedema**
1994. **Erythema multiforme (minor & major)**
1995. **EMPACT syndrome (Erythema multiforme associated with Phenytoin And Cranial radiation Therapy)**
1996. **Erythema nodosum EN**
1997. **Erythema nodosum Like lesion in Behcet disease**
1998. **Erythema nodosum leprosum**
1999. **Erythema nodosum migrans**
2000. **Erythema nuchae (salmon patch)**
2001. **Erythema palmare hereditarium**
2002. **Erythema papulosa semicircularis recidivans = Erythema papulatum centrifugum**
2003. **Erythema punctatum Higuchi (EPH)**
2004. **Erythema rhematicum migrans**
2005. **Concentric centrifugal figurate erythema**
2006. **Palpable migratory arciform erythema**
2007. **Erythema scarlatiniforme desquamativum recidivans**
2008. **Erythema toxicum neonatorum =Toxic erythema of the newborn**
2009. **Pustular erythema toxicum neonatorum**

2010. **Erythrasma**
2011. **Disciform erythrasma**
2012. **Erythrocyanosis**
2013. **Erythroderma**
2014. **Idiopathic erythroderma**
2015. **Erythrodermic trichophyton**
2016. **Erythrokeratolysis Hiemalis =keratolytic winter erythema=Oudtshoorn disease**
2017. **Erythroleukoplakia**
2018. **Erythroplakia**
2019. **Erythromelalgia= Erythralgia= Erythermalgia**
2020. **Erythromelanosis follicularis faciei et colli**
2021. **Erythroplasia of Queyrat**
2022. **Erythrose péribuccale of brocq**
2023. **Erythrosis pigmentosa mediofacialis**
2024. **Erasmus syndrome**
2025. **Essential fatty acid deficiency**
2026. **Estrogen dermatitis**
2027. **Ewing's sarcoma= peripheral primitive neuroectodermal tumor/PNET**
2028. **Evans syndrome**
2029. **Exserohilum infection**
2030. **Excited skin syndrome**
2031. **Exercise-induced vasculitis =Golfer vasculitis**
2032. **Exfoliative dermatitis**
2033. **Extramedullary hematopoiesis**
2034. **Extravasation injuries**
2035. **Fabricated&induced illness=Munchhausen syndrome by proxy**
2036. **Fabry disease**
2037. **FACE Syndrome : facial-afro–caribbean- childhood eruption**
2038. **Facial dermal dysplasia**
2039. **Facial discoid dermatosis FDD**
2040. **Facial idiopathic granulomas with regressive evolution FIGURE**
2041. **Facial overfilled syndrome**

2042. **Facial papules of frontal fibrosing alopecia**
2043. **Facio-auriculovertebral syndrome**
2044. **Facio-cutaneous-skeletal syndrome**
2045. **Facio-digito-genital syndrome**
2046. **Facio-genital popliteal syndrome**

Facies in dermatology (see next)

2047. ✓ **Acrogeric facies**
2048. ✓ **Acromegaloid facies**
2049. ✓ **Adenoid facies**
2050. ✓ **Amiodarone facies**
2051. ✓ **Angelic facies**
2052. ✓ **Antonine facies**
2053. ✓ **Asiatic porcelain doll facies**
2054. ✓ **Atopic facies**
2055. ✓ **Bird-like facies**
2056. ✓ **Bloodhound facies**
2057. ✓ **Bull dog face**
2058. ✓ **Cadaveric facies**
2059. ✓ **Cigarette facies**
2060. ✓ **Coarse facies**
2061. ✓ **Dengue facies**
2062. ✓ **Dog=simian facies**
2063. ✓ **Drawn facies**
2064. ✓ **Dysmorphic facies**
2065. ✓ **Elfin facies**
2066. ✓ **Gargoyles facies**
2067. ✓ **Gaunt facies**
2068. ✓ **Greek helmet facies**
2069. ✓ **Grimace-like facies**
2070. ✓ **Hippocratic facies**
2071. ✓ **Latrodectus facies**

2072. ✓ **Leonine facies**
2073. ✓ **Mask like face**
2074. ✓ **Mickey mouse facies**
2075. ✓ **Mitral face**
2076. ✓ **Mongoloid face=flat facies**
2077. ✓ **Monkey facies**
2078. ✓ **Moon facies**
2079. ✓ **Old man facies**
2080. ✓ **Sad man facies**
2081. ✓ **Slapped cheek facies**
2082. ✓ **Tabetic facies**
2083. ✓ **Triangular face**
2084. ✓ **Whistling facies**
2085. **Failed scalp micropigmentation**
2086. **Familial annular erythema**
2087. **Familial atypical mole syndrome**
2088. **Familial cerebral cavernous malformation**
2089. **Familial cold auto-inflammatory syndrome FCAS**
2090. **Familial cutaneous and mucosal venous malformation**
2091. **Familial dysautonomia**
2092. **Familial dyskeratotic comedon**
2093. **Familial disseminated comedones without dyskeratosis (FDCWD)**
2094. **Familial flame nevi**
2095. **Familial gigantic melanocytosis**
2096. **Familial haemophagocytic lymphohistiocytosis**
2097. **Familial histiocytic dermatoarthritis**
2098. **Familial mediterranean fever (FMF)**
2099. **Familial multiple acral mucinous fibrokeratomas**
2100. **Familial multiple café-au-lait spots**
2101. **Familial multiple discoid fibromas (FMDF)= Familial Multiple Trichodiscomas**
2102. **Familial multiple lipomatosis**

2103. **Familial primary cutaneous amyloidosis**
2104. **Familial progressive hyperpigmentation**
2105. **Familial progressive hypo-and hyperpigmentation**
2106. **Familial speckled acral hypopigmentation**
2107. **Factor V Leiden mutation**
2108. **Fanconi anemia**
2109. **Far Eastern spotted fever**
2110. **Farber lipogranulomatosis disease= Ceramidase deficiency**
2111. **Fascioliasis**
2112. **Fat-storing hamartoma of dermal dendrocytes**
2113. **Faun tail= Lumbosacral Hypertrichosis**
2114. **Fegeler syndrome(acquired capillary malformation)**
2115. **Felty's syndrome**
2116. **Female genital mutilation**
2117. **Ferguson–Smith syndrome**
2118. **Ferro-Cerebro-Cutaneous (FCC) syndrome = Neurodevelopmental Disorder with Epilepsy and Hemochromatosis (NEDEPH)**
2119. **Fetal alcohol syndrome**
2120. **Fetal hydantoin syndrome**
2121. **FG syndrome =Opitz-Kaveggia syndrome**
2122. **Fiberglass dermatitis**
2123. **Fibroadenoma**
2124. **Fibro adipose vascular anomaly FAVA**
2125. **Fibroblastic connective tissue nevus FCTN**
2126. **fibroblastic rheumatism**
2127. **Fibroelastolytic papulosis of neck**
2128. **Fibroblastoma**
2129. **Desmoplastic Fibroblastoma =Collagenous Fibroma**
2130. **Dermal desmoplastic fibroblastoma**
2131. **Fibroepithelial polyp**
2132. **Fibroepithelioma of pinkus**
2133. **Fibrolipoma**
2134. **Liposurfomas(Acquired fibrolipomas)**

2135. **Fibrolipomatous hamartoma**
2136. **Hemosiderotic fibrolipomatous tumor**
2137. **Fibrofolliculoma**
2138. **Fibrokeratoma**
2139. **Fibroma**
2140. **Fibroma of tendon sheath**
2141. **Cementifying fibroma =ossifying fibroma**
2142. **Fibroma of oral mucous membrane =bite fibroma= Focal fibrous hyperplasia**
2143. **Fibroma fissuratum**
2144. **Fibromatosis colli**
2145. **Fibromatoses (see next)**
2146. ✓ **Desmoid tumor**
2147. ✓ **knuckle pads (holoderma)**
2148. ✓ **pachydermodactyly**
2149. ✓ **palmar fibromatosis (Dupuytren disease)**
2150. ✓ **penile fibromatosis (Peyronie disease)**
2151. ✓ **plantar fibromatosis (Ledderhose disease)**
2152. ✓ **superficial fascial fibromatosis**
2153. **Fibrodysplasia ossificans progressive**
2154. **Fibroelastolytic papulosis**
2155. **Fibro-osseous pseudotumor of the digits FOPT**
2156. **Fibrosarcoma (see next)**
2157. ✓ **Classic fibrosarcoma**
2158. ✓ **Fibromyxoid fibrosarcoma**
2159. ✓ **Myxofibrosarcoma&malignant fibrous histiocytoma**
2160. ✓ **Sclerosing epithelioid fibrosarcoma**
2161. ✓ **Congenital=Infantile Fibrosarcoma**
2162. ✓ **Adult-type fibrosarcoma**
2163. **Fibrous hamartoma of infancy**
2164. **Fibrous papule of nose**
2165. **Fibrous papule of lips**

2166. **Fiddler's neck**
2167. **Finlay–Marks syndrome, scalp–ear–nipple syndrome**
2168. **Finger pebbles**
2169. **Fish odour syndrome**
2170. **Fisher-volavsek syndrome**
2171. **Fisch syndrome**
2172. **Fitzsimmons-McLachlan-Gilbert syndrome**
2173. **Fixed drug eruption FDE**
2174. **FDE variants (Nonpigmenting&Inverse &Linear& Wandering&Chronic& Cellulitis like& Psoriasiform& Neutrophilic&Erythema dyschromicum perstans like&FDE with pigmentary loss&generalized bullous &linear&Paronychia)**
2175. **Fixed eczematous eruption**
2176. **Fixed food eruption**
2177. **Fixed sunlight eruption**

Flagellate dermatoses (see next)

2178. ✓ **adult-onset still's disease**
2179. ✓ **bendamustine**
2180. ✓ **bleomycin**
2181. ✓ **chikungunya fever induced flagellate pigmentation**
2182. ✓ **child & partner &elder abuse**
2183. ✓ **cnidarian (portuguese man-of-war and jellyfish) stings**
2184. ✓ **dermatitis artefacta**
2185. ✓ **dermatographism**
2186. ✓ **dermatomyositis**
2187. ✓ **docetaxel**
2188. ✓ **excoriations by pruritic conditions**
2189. ✓ **hypereosinophilic syndrome**
2190. ✓ **idiopathic flagellate pigmentation**
2191. ✓ **paederus and other insects**
2192. ✓ **peplomycin**
2193. ✓ **phytophotodermatitis**

2194. ✓ poison ivy dermatitis
2195. ✓ religious punishment
2196. ✓ sexual pleasure (sadomasochism)
2197. ✓ shiitake mushroom ingestion (raw or undercooked)
2198. ✓ torture
2199. Flagellate hyperpigmentation
2200. Flagellate hypopigmentation
2201. Flag-like hypomelanosis
2202. Flag-like hypermelanosis
2203. Flag-like lentiginosis
2204. Flea-borne spotted fever
2205. Flea-borne typhus=California flea rickettsiosis
2206. Flegel's diseases= hyperkeratosis lenticularis perstans
2207. Flinders Island spotted fever
2208. Floppy eyelid syndrome
2209. Flotch syndrome
2210. Florid papillomatosis
2211. Florid reactive periostitis
2212. Flushing
2213. Flynn-aird syndrome
2214. Focal facial dermal dysplasia (FFDD)
2215. ✓ ffdd1 – Brauer
2216. ✓ ffdd2 – Brauer–Setleis
2217. ✓ ffdd3 – Setleis
2218. ✓ ffdd4 – preauricular
2219. Focal epithelial hyperplasia
2220. Focal Fibrous Hyperplasia
2221. Folliculosebaceous cystic hamartoma (FSCH)= sebofolliculoma
2222. Follicular keratosis
2223. Follicular occlusion triad
2224. Follicular occlusion tetrade
2225. Actinic superficial folliculitis ASF

2226. **Actinic folliculitis**
2227. **Candidal folliculitis**
2228. **Demodex folliculitis**
2229. **Dissecting terminal hair folliculitis**
2230. **Disseminate and recurrent infundibulofolliculitis (DRIF)= Hitch and Lund disease**
2231. **Eosinophilic pustular folliculitis of infancy**
2232. **Eosonophilic folliculitis of ofuji**
2233. **Eosinophilic folliculitis associated with HIV infection**
2234. **Immunosuppression-associated eosinophilic pustular folliculitis**
2235. **Eosinophilic pustular folliculitis**
2236. **Folliculitis Decalvans = Quinquaud's Disease= Acne Decalvans**
2237. **Folliculitis Decalvans-Like Pustular Plaques on the Limbs**
2238. **Folliculitis nares perforans**
2239. **Friction folliculitis**
2240. **Gram negative folliculitis**
2241. **Hemorrhagic folliculitis in scurvy**
2242. **Herpes simplex folliculitis**
2243. **Hot tub folliculitis**
2244. **Irritation folliculitis**
2245. **Perforating folliculitis**
2246. **Pityrosporum folliculitis= Malassezia folliculitis**
2247. **Pseudolymphoma folliculitis**
2248. **Folliculitis spinulosa decalvans**
2249. **Sterile neutrophilic folliculitis with perifollicular vasculopathy**
2250. **Superficial pustular folliculitis=Impetigo of Bochart**
2251. **Superficial pustular folliculitis of the face**
2252. **Traumatic anserine folliculosis**
2253. **Trunk folliculitis**
2254. **Folliculocystic and collagenous hamartoma**
2255. **Fordyce spots =Ectopic sebaceous glands**
2256. **Foreign body granuloma**
2257. **Formication**

2258. **Fox-Fordyce disease =Apocrine miliaria**
2259. **Fournier gangrene**
2260. **François syndrome**
2261. **Frank Ter Haar syndrome**
2262. **Franceschetti-klein syndrome= Mandibulofacial dysostosis**
2263. **Freckles = Ephelid**
2264. **Frey syndrome**
2265. **Fryns syndrome**
2266. **Friction amyloidosis**
2267. **Frictional asymptomatic darkening of the extensor surfaces (FADES)**
2268. **Frictional hypermelanosis**
2269. **Frictional Keratosis**
2270. **Frictional lichenoid dermatitis**
2271. **Frictional dermal melanosis=Lifa disease**
2272. **Frictional sweat dermatitis**
2273. **Fried tooth and nail syndrome**
2274. **Frontometaphyseal dysplasia**
2275. **Frost bite**
2276. **Fucosidosis**
2277. **Fuhrmann syndrome**
2278. **Furuncle**
2279. **Fusariosis=Hyalohyphomycosis**
2280. **Futcher's lines**
2281. **Galli-Galli diseas**
2282. **Gamasoidosis**
2283. **Galactosialidosis**
2284. **Ganastomiasis**
2285. **Ganglion cyst**
2286. **Gangliosidosis**
2287. **Ganglioneuroma**
2288. **Gangrene**
2289. **GAPO syndrome (growth retardation, alopecia, pseudoanodontia and optic atrophy)**

2290. **Gardner syndrome= Familial polyposis of the colon**
2291. **Gardner-associated fibroma**
2292. **Gardner-Diamond syndrome=Autoerythrocyte sensitization**
2293. **Gas gangrene**
2294. **Gastrocutaneous syndrome**
2295. **Gata2 deficiency/ MonoMAC syndrome**
2296. **Gaucher disease/Sphingolipidoses**
2297. **Gazali-temple syndrome**
2298. **Gemcitabine-induced pseudocellulitis**
2299. **GEMSS Syndrome(glaucoma, lens ectopia, microspherophakia, joint stiffness and stocky short "pseudoathletic" build)**
2300. **Genralized essential telangiectasia**
2301. **Generalised exanthematous pustular dermatophytids**
2302. **Generalized pustular figurate erythema**
2303. **Generalized verrucosis (GV)**
2304. **Genodermatoses**
2305. **Geographical flushing of the children's face**
2306. **Geotrichosis**
2307. **Geroderma osteodysplastica**
2308. **Gianotti-Crosti syndrome =papular acrodermatitis of childhood= Infantile papular acrodermatitis=papulovesicular acrolocated syndrome**
2309. **Giant axonal neuropathy with curly hair**
2310. **Giant cell arteritis =Temporal arteritis**
2311. **Giant cell angioblastoma**
2312. **Giant cell fibroma**
2313. **Giant cell granuloma**
2314. **Giant cell lichenoid dermatitis**
2315. **Giant cell tumor tendon sheath= Tenosynovial Giant Cell Tumor**
2316. **Giant Cell Tumor of Soft Tissue**
2317. **Gingival cyst of the adult**
2318. **Gingival cyst of the newborn**
2319. **Gingival fibromatosis**
2320. **Gigantic melanocytosis**

2321. **Gigantomastia**
2322. **Gingival fibromatosis and hypertrichosis**
2323. **Gingival fibromatosis –sparse hair –malposition of teeth**
2324. **Gingival hyperplasia**
2325. **Drug-induced gingival overgrowth**
2326. **Desquamative Gingivitis**
2327. **Necrotizing ulcerative gingivitis**
2328. **Glanders**
2329. **Glandular cheilitis**
2330. **Glanzmann–Riniker syndrome**
2331. **Gleich syndrome**
2332. **Nasal Glioma=Nasal cerebral heterotopia**
2333. **Glomeruloid hemangioma**
2334. **Glomangiopericytoma**
2335. **Glomus tumour**
2336. **Glomangioma**
2337. **Glomangiomyoma**
2338. **Glomangiomatosis**
2339. **Glomangiosarcoma**
2340. **Glomuvenous malformation**
2341. **Glossitis**
2342. **Atrophic glossitis**
2343. **Hunter glossitis**
2344. **Median rhomboid glossitis=Central papillary atrophy**
2345. **Glossodynia**
2346. **Glucagonoma syndrome**
2347. **Glutaric aciduria**
2348. **Glycogen storage disease**
2349. **Gnas mutation disorders**
2350. **Gnathostomiasis=Nodular migratory eosinophilic panniculitis**
2351. **Gnathophyma**
2352. **Gobello Naevus syndrome**
2353. **Goeminne syndrome**

2354. **Goitre**
2355. **Goldenhar syndrome=Oculo-auriculo-Vertebral Dysplasia**
2356. **Goltz -Gorlin syndrome=Focal dermal hypoplasia= Goltz syndrome**
2357. **Gomez-lopez-hernandez syndrome= Cerebello-trigemino-dermal**
2358. **Gonorrhoea**
2359. **Gonococcemia**
2360. **Gordonia bronchialis**
2361. **Gorlin-Goltz syndrome**
2362. **Gorlin–Chaudry–Moss syndrome**
2363. **Good syndrome**
2364. **Gorham-Stout syndrome**
2365. **Goodpasture syndrome**
2366. **✓Gottron papule**
2367. **✓Inverse Gottron papule**
2368. **✓Sclerodermoid amyloidosum gottron**
2369. **✓Gottron sign**
2370. **✓Gottron-like papule**
2371. **✓Gottron acrogeria (Gottron syndrome)**
2372. **✓Papillomatosis cutis carcinoides of Gottron**
2373. **✓Darrier-Gottron erythrokeratoderma=progressive symmetric erythrokeratoderma**
2374. **Gout = Arthritis urica &tophi**
2375. **Gouty panniculitis**
2376. **Pseudo Gout**
2377. **Granddad syndrome**
2378. **Graft versus host disease GVHD (Acute&Chronic)**
2379. **Graham-Little-piccardi-Lasseur syndrome**
2380. **Grönblad-strandberg syndrome**
2381. **Granular cell tumour =Abrikossoff's tumor**
2382. **Granular parakeratosis GP** **(see next)**
2383. **✓Follicular granular parakeratosis**
2384. **✓Granular parakeratosis of eccrine ostia**

2385. ✓ Granular parakeratotic acanthoma
2386. ✓Incidental GP
2387. ✓ Intertriginous GP
2388. Granular parakeratotic acanthoma
2389. Granulosis rubra nasi
2390. Granulocytic spongiotic papulovesiculosis
2391. Granuloma annulare **(see next)**
2392. ✓Localized
2393. ✓Generalized
2394. ✓Perforating
2395. ✓Subcutaneous=deep
2396. ✓Disseminated
2397. ✓Patch
2398. ✓ Photodistribute
2399. ✓Follicular pustular variant
2400. ✓Targetoid
2401. ✓Giant
2402. ✓Arcuate dermal erythema
2403. ✓Anetodermic
2404. ✓Acute-onset painful acral
2405. ✓Interstitial
2406. ✓papular
2407. ✓linear
2408. Granuloma faciale
2409. Extrafacial granuloma faciale
2410. Granuloma fissuratum
2411. Granuloma gluteal infantum
2412. Granuloma inguinale
2413. Granuloma multiform
2414. Granulomatous blepharitis
2415. Granulomatous cheilitis

2416. **Granulomatous slack skin**
2417. **Granulomatous vasculitis**
2418. **Gregg syndrome**
2419. **Greither's syndrome**
2420. **Grey baby syndrome**
2421. **Griscelli –Prunieras syndrome (3 types)**
2422. **Grover disease= Transient Acantholytic Dermatosis**
2423. **Grybowski type generalized eruptive keratoacanthoma**
2424. **Guinea worm disease**
2425. **Gynecomastia**
2426. **H syndrome**
2427. **Haber's syndrome**
2428. **Haberland syndrome**
2429. **Hajdu-Cheney syndrome**
2430. **Haim-Munk syndrome**
2431. **Hair-thread tourniquet syndrome (HTTS)**
2432. **Halo nevus**
2433. **HAIR-AN syndrome hyperandrogenemia, insulin resistance, acanthosis nigricans**
2434. **Haemangioma = Naevus Vasculosus=Angiomatous Nevi**
2435. **Diffuse Neonatal Hemangiomatosis =Miliary Hemangiomatosis =Disseminated Hemangiomatosis**
2436. **LUMBAR syndrome (lower-body hemangioma& urogenital abnormalities=ulceration&myelopathy (severe compression) of the spinal cord&bony deformities&anorectal malformations and arterial anomalies& renal anomalies)**
2437. **PHACES syndrome (p = posterior fossa (this refers to possible abnormal structures in the brain, particularly the cerebellum.)&h = hemangioma&a = arterial (this refers to possible abnormal arteries in the brain, neck or chest.)&c = cardiac (this refers to possible heart defects or abnormalities of the large blood vessels connected to the heart.)&e = eyes (this refers to possible eye or vision issues&s = sternal cleft)**

2438. **PELVIS syndrome (perineal haemangioma&external genitalia malformations & lipomyelomeningocele&vesicorenal abnormalities&imperforate anus& skin tag)**
2439. **Noninvoluting congenital haemangioma NICH**
2440. **Rapidly involuting congenital haemangioma RICH**
2441. **Partially involuting congenital haemangioma PICH**
2442. **Multifocal infantile haemangiomas**
2443. **Haemangiopericytoma**
2444. **Haemochromatosis**
2445. **Hailey -Hailey disease= Familial benign chronic pemphigus**
2446. **Hair cast=pseudonits**
2447. **Hair-follicle nevus**
2448. **Hair heterochromia**
2449. **Hair matrix cyst**
2450. **Red hair=Rutilism**
2451. **Hair transplant related diseases**
2452. **Hair Weathering**
2453. **Hair Photoaging**
2454. **Halo dermatitis**
2455. **Halo scalp ring**
2456. **Halogenoderma**
2457. **Hallermann–Streiff syndrome=François Dyscephalic syndrome=Oculomandibulodyscephaly with Hypotrichosis**
2458. **Hand-Foot-and-Mouth disease HFMD**
2459. **Pseudomonas hand foot syndrome**
2460. **Hand foot skin reaction**
2461. **Hand foot skin syndrome**
2462. **Hand–Foot–Genital syndrome**
2463. **Haploinsufficiency of A20**
2464. **Happle–tinschert syndrome**
2465. **Harlequin colour change of newborn**
2466. **Harlequin syndrome**
2467. **Hartnup disease**

2468. **Hay-Wells syndrome**
2469. **Hayden syndrome**
2470. **Heamatohidrosis**
2471. **Heamngiopericytoma**
2472. **Hemolytic uremic syndrome**
2473. **Heart hand syndrome**
2474. **Heat cramps**
2475. **Heat edema**
2476. **Heat stroke**
2477. **Heat syncope**
2478. **Heat-related illnesses**
2479. **Heberden nodule**
2480. **Heck disease=Focal epithelial hyperplasia**
2481. **Black heel =Talon noir-=Calcaneal petechiae**
2482. **Heel-line hyperpigmentation**
2483. **Heel fissures**
2484. **Heel stick injury**
2485. **Heel stick calcinosis**
2486. **Hematoma**
2487. **Hemifacial atrophy**
2488. **Hemifacial mixed appendageal tumor**
2489. **Hemihyperplasia lipomatosis**
2490. **Hemi-maxillo-facial dysplasia**
2491. **Hemochromatosis**
2492. **Hemorrhagic insect bites**
2493. **Hemosiderotic fibrolipomatous tumor (HFLT)**
2494. **Hemophagocytic syndrome**
2495. **Henna allergic dermatitis**
2496. **Hennekam lymphangiectasia–lymphedema syndrome**
2497. **Henoch schonlein purpura= anaphylactoid purpura**
2498. **Heparin-induced thrombocytopenia syndrome (HITS)=Heparin-associated thrombocytopenia with thrombosis (HATT) syndrome**

2499. **He remained (Hematologic related malignancy induced eosinophilic dermatoses)**
2500. **Hereditary acrokeratotic poikiloderma=Weary syndrome=Dowling syndrome**
2501. **Hereditary alpha tryptasemia**
2502. **Hereditary Angioedema**
2503. **Hereditary benign intraepithelial dyskeratosis**
2504. **Hereditary benign telangiectasia**
2505. **Hereditary congenital hypopigmented and hyperpigmented macules (westerhof syndrome)**
2506. **Hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP)**
2507. **Hereditary generalized punctate leukoderma**
2508. **Hereditary gingival fibromatosis**
2509. **Hereditary hemorrhagic telangiectasia =osler-weber-rendu syndrome**
2510. **Hereditary leiomyomatosis and renal cell cancer (HLRCC)**
2511. **Hereditary mucoepithelial dysplasia**
2512. **Hereditary multiple exostosis (diaphyseal aclasis)**
2513. **Hereditary multiple lipomas**
2514. **Hereditary neurocutaneous angiomatosis**
2515. **Hereditary nonsyndromic multiple basal cell carcinoma**
2516. **Hereditary onycho-osteodysplasia (HOOD)**
2517. **Hereditary osteo-onychodysplasia**
2518. **Hereditary papulotranslucent acrokeratoderma**
2519. **Hereditary premature canities**
2520. **Hereditary sclerosing poikiloderma of weary**
2521. **Hereditary sensory and autonomic neuropathies (HSAN):(IA&IB&IC&II&III&IV& V&HSAN with spastic paraplegia)**
2522. **Heerfordt-Waldenström syndrome**
2523. **Hermansky Pudlak syndrome (9 types)**
2524. **Herpangina**
2525. **Herpes simplex (orolabial& gingivostomatitis & genital & gladiatorum & folliculitis & encephalitis & proctitis & whitlow)**

2526. **Herpes simplex (Neonatal& Intrauterine)**
2527. **Herpetic geometric glossitis**
2528. **Herpes vegetans**
2529. **Herpes zoster=shingles (See next)**
2530. **✓Classic**
2531. **✓Abortive**
2532. **✓Hemorrhagic**
2533. **✓Bullous**
2534. **✓Gangrenous**
2535. **✓Disseminated**
2536. **✓Zoster sine herpes**
2537. **✓ Duplex symmetricus**
2538. **✓Duplex bilateralis**
2539. **✓Duplex unilateralis**
2540. **✓Herpetic zoster folliculitis**
2541. **Heterochromia**
2542. **Heterotopic neural tissue of the skin**
2543. **Heterotopic salivary gland tissue**
2544. **Hibernoma**
2545. **Hidroacanthoma simplex=Intraepidermal Poroma**
2546. **Malignant hidroacanthoma simplex**
2547. **Neutrophilic eccrine hidradenitis NEH**
2548. **Idiopathic/recurrent palmoplantar hidradenitis**
2549. **Idiopathic plantar hidradenitis (IPH)**
2550. **Hidradenitis suppurativa HS=acne inversa=Verneuil's disease**
2551. **Hidradenoma**
2552. **Apocrine hidradenoma**
2553. **Hidradenoma papilliferum**
2554. **Poroid Hidradenoma**
2555. **Hidradnocarcinoma**
2556. **Hidradenocarcinoma papilliferum**
2557. **Hidrocystoma (apocrine& eccrine)**

2558. **Solitary (smith) hidrocystoma**
2559. **Hirsutism**

Histiocytosis

2560. **LCH langerhan cell histiocytosis(see next)**
2561. **Adult type langerhan cell histiocytosis**
2562. ✓ **Letterer-Siwe disease**
2563. ✓ **Hand-Schüller-Christian disease**
2564. ✓ **Congenital self-healing reticulohistiocytosis (CSHR) =Hashimoto-Pritzker disease**
2565. ✓ **Eosinophilic granuloma**
2566. **Non LCH langerhan cell histiocytosis =Non-X histiocytosis=Class II histiocytosis (see next)**
2567. ✓ **Progressive nodular histiocytosis PNH= Spindle Cell Xanthogranuloma**
2568. ✓ **Hereditary Progresive mucinosis histiocytosis**
2569. ✓ **Papular xanthoma**
2570. ✓ **Generalized eruptive histiocytosis GEH / histiocytoma GEH**
2571. ✓ **Benign cephalic histiocytosis= Papular Histiocytoses of The Head**
2572. ✓ **Indeterminate histiocytosis**
2573. ✓ **S100-negative, CD1a-positive histiocytosis**
2574. ✓ **Giant cell reticulohistiocytoma**
2575. ✓ **Solitary reticulohistiocytoma= Solitary Epithelioid Histiocytoma**
2576. ✓ **Adult xanthogranuloma**
2577. ✓ **Juvenile xanthogranuloma**
2578. ✓ **Symmetrical giant facial plaque-type juvenile xanthogranuloma (SGFP-JXG)**
2579. ✓ **Necrobiotic xanthogranuloma**
2580. ✓ **Xanthoma disseminatum XD=Montgomery syndrome=Disseminated xanthosiderohistiocytosis (a variant)**
2581. ✓ **Multicentric reticulohistiocytosis**
2582. ✓ **Erdheim-chester disease= Uber Lipoid Granulomatosis)**
2583. ✓ **Kikuchi-Fujimoto Disease= Histiocytic necrotizing lymphadenitis**

2584. ✓ Familial Sea blue histiocytosis
2585. ✓ Hemophagocytic lymphohistiocytosis
2586. ✓ Rosai–Dorfman disease RDD =Sinus histiocytosis with massive lymphadenopathy SHML
2587. **Malignant Histiocytosis=Class III histiocytosis (see next)**
2588. ✓ Histiocytic sarcoma
2589. ✓ Langerhans cell sarcoma
2590. ✓ Follicular dendritic cell sarcoma
2591. ✓ Interdigitating dendritic cell sarcoma
2592. ✓ True histiocytic lymphoma= reticulum cell sarcoma=histiosarcoma
2593. crystal-storing histiocytosis
2594. Histoplasmosis= Darling's Disease= Ohio Valley Disease
2595. Hitch syndrome (hearing impairment, undescended testis, circumferential skin creases, and mental handicap)
2596. Hobnail hemangioma=Targetoid hemosiderotic lymphatic malformation=Targetoid Hemosiderotic Hemangioma= Hobnail Lymphatic Malformation
2597. Hobnail hemangioendothelioma family (retiform hemangioendothelioma and papillary intralymphatic angioendothelioma)
2598. Holocarboxylase synthetase deficiency
2599. Holt-oram syndrome
2600. Homocystinuria
2601. Hooft disease
2602. Hookworm disease
2603. Hope disease
2604. HOPP (hypotrichosis, acro-osteolysis=onychogryphosis, palmoplantar keratoderma, and periodontitis) syndrome
2605. Horizontal forehead lines
2606. Hornstein-knickenberg syndrome
2607. Hoyeraal–hreidarrson syndrome
2608. Human monocytotropic ehrlichioses

2609. **Human granulocytic anaplasmosis**
2610. **Ehrlichia muris-like agent ehrlichiosis**
2611. **Hunan hand syndrome**
2612. **Huntley's papule**
2613. **✓Hutchinson's sign**
2614. **✓Hutchinson's triad**
2615. **✓Hutchinson's teeth**
2616. **✓Hutchinson melanotic freckle**
2617. **✓Hutchinson's melanotic disease**
2618. **✓Hutchinson–Gilford progeria syndrome**
2619. **✓Hutchinson's facies**
2620. **✓Hutchinson's angina**
2621. **✓Hutchinson–Horton syndrome**
2622. **✓Hutchinson's prurigo**
2623. **✓Hutchinson's patch**
2624. **✓Hutchinson's dyshidrosis**
2625. **Hyalohyphomycosis**
2626. **Hybrid cysts**
2627. **Hydatid cyst/Echinococcosis**
2628. **Hydroa vacciniforme lymphoproliferative disorder**
2629. **Hydroa vacciniforme=Bazin's hydroa vacciniforme**
2630. **Hydroa vacciniforme-like cutaneous t-cell lymphoma**
2631. **Hydroxyurea-induced ulcers**
2632. **4-hydroxybutyric aciduria**
2633. **Hyper eosinophilic syndrome HES**
2634. **Hypereosinophilic dermatitis of Nir-Westfried**
2635. **Hyperirritable skin**
2636. **Hyper IgD syndrome**
2637. **Hyper IgE syndrome=Job syndrome HIES**
2638. **Hyper linearity palm -sole**
2639. **Hyperkeratotic flexural erythema**
2640. **Hyperkeratotic cutaneous capillary–venous malformation**

2641. **Hypertrophic scar**
2642. **Hyperhidrosis**
2643. **Hyperpigmented macules on the face of young children (HMFYC)**
2644. **Hypertrichosis (see next)**
2645. **✓Congenital hypertrichosis lanuginose**
2646. **✓X-linked hypertrichosis (congenital generalized hypertrichosis)**
2647. **✓Acquired generalized hypertrichosis**
2648. **✓Acquired hypertrichosis lanuginosa**
2649. **✓universal hypertrichosis**
2650. **✓Ambras syndrome**
2651. **✓x-linked hypertrichosis**
2652. **✓Hypertrichosis of the auricle**
2653. **✓Hypertrichosis of the nasal tip**
2654. **✓Trichomegaly of the eyelashes**
2655. **✓Hypertrichosis of the eyebrows**
2656. **✓prepubertal hypertrichosis**
2657. **✓Hypertrichosis cubiti**
2658. **✓Anterior cervical hypertrichosis**
2659. **✓Posterior cervical hypertrichosis**
2660. **✓Hairy palms and soles**
2661. **✓Hairy ear pinna**
2662. **✓Hairy elbows syndrome**
2663. **✓Gingival fibromatosis with hypertrichosis**
2664. **✓Acromegaly with hypertrichosis**
2665. **Hypertrophy of circumvallate papillae**
2666. **Hypertrophy of foliate papillae**
2667. **Hypertrophy of the fungiform papillae**
2668. **Hypervitaminosis A**
2669. **Hypervitaminosis B (B3,B6)**
2670. **Hypervitaminosis D**
2671. **Hypervitaminosis E**

2672. **Hypodermatitis sclerodermaformis**
2673. **Hypodontia and nail dysgenesis**
2674. **Hypoglossia-Hypodactylia anomaly**
2675. **Hypohidrosis**
2676. **Hypohidrosis with diabetes insipidus= fleck's syndrome**
2677. **Hypohidrosis with neurolabyrinthitis**
2678. **Hypomelia–Hypotrichosis–facial hemangioma syndrome =pseudothalidomide syndrome**
2679. **Hypophosphatasia (HPP)**
2680. **Hypopigmented scar**
2681. **Hypothenar hammer syndrome**
2682. **Hypotrichosis (see next)**
2683. ✓ **Hypotrichosis, Marie**
2684. ✓ **Hypotrichosis, Marie Unna**
2685. ✓ **Hypotrichosis simplex**
2686. ✓ **Hypotrichosis simplex of the scalp**
2687. ✓ **Hypotrichosis with juvenile macular dystrophy**
2688. ✓ **Hypotrichosis with recurrent skin "vesicles"**
2689. ✓ **Localized autosomal recessive hypotrichosis**
2690. ✓ **Hypotrichosis–lymphedema–telangiectasia syndrome**
2691. **Ichthyosis (see next)= Fish-skin disease=alligator skin**
2692. ✓ **Ichthyosis vulgaris=Ichthyosis simplex**
2693. ✓ **Lamellar Ichthyosis =Lamellar exfoliation of the newborn**
2694. ✓ **Autosomal dominant Lamellar Ichthyosis**
2695. ✓ **X linked recessive Ichthyosis = Steroid sulfatase deficiency**
2696. ✓ **Collodion baby/ Congenital self-healing collodion baby**
2697. ✓ **Harlequin ichthyosis = ichthyosis congenita gravior =Harlequin fetus**
2698. ✓ **Bullous congenital ichthyosiform erythroderma BCIE=Epidermolytic ichthyosis of brocq=Bullous Ichthyosis**
2699. ✓ **Cyclic ichthyosis with epidermolytic hyperkeratosis**

2700. ✓Congenital ichthyosiform erythroderma= Non-bullous congenital ichthyosiform erythroderma NBCIE= Ichthyosis Congenita Type 1
2701. ✓Annular epidermolytic ichthyosis
2702. ✓Mosaic epidermolytic ichthyosis=Epidermolytic type of epidermal nevus
2703. ✓Autosomal recessive exfoliative ichthyosis
2704. ✓Autosomal recessive congenital ichthyosis ARCI
2705. ✓Autosomal recessive ichthyosis with hypotrichosis (ARIH)
2706. ✓Autosomal recessive keratoderma-ichthyosis-deafness syndrome ARKID
2707. ✓Chanarin–Dorfman syndrome =Neutral lipid storage disease with ichthyosis
2708. ✓CHILD syndrome: congenital hemidysplasia with ichthyosiform erythroderma and limb defects
2709. CHIME(Colobomas, Heart defect, Ichthyosiform dermatosis, Mental retardation, Ear anomalies)= Zunich neuroectodermal syndrome
2710. ✓Comel -Netherton syndrome= Ichthyosis linearis circumflexa
2711. ✓Conradi–Hünermann–Happle syndrome= X-linked dominant chondrodysplasia punctata type 2
2712. Rhizomelic chondrodysplasia punctate (autosomal recessive chondrodysplasia punctata type 1)
2713. ✓ Bathing suit Ichthyosis
2714. ✓Ichthyosis bullosa of Siemens =superficial epidermolytic ichthyosis =Ichthyosis exfoliativa
2715. ✓Ichthyosis cribriformis
2716. ✓Ichthyosis en confetti=Confetti Ichthyosis=Congenital Reticular Ichthyosiform Erythroderma CRIE =Ichthyosis with Confetti= Ichthyosis Variegata
2717. ✓Ichthyosis follicularis with atrichia and photophobia IFAP
2718. ✓Ichthyosis hystrix= ichthyosis hystrix curth-macklin=porcupine man=systematized verrucous nevus
2719. ✓HID: Hystrix-like ichthyosis with deafnes
2720. ✓Ichthyosis prematurity syndrome IPS

2721. ✓Ichthyosis with hypogonadism
2722. ✓Ichthyosis, spastic quadriplegia, mental retardation ISQMR
2723. ✓Ichthyosis-hypotrichosis-sclerosing cholangitis syndrome (IHSC) syndrome
2724. ✓Gaucher syndrome type 2
2725. ✓Ichthyosis–Hypotrichosis syndrome
2726. ✓Neonatal ichthyosis–sclerosing cholangitis (NISCH) syndrome
2727. ✓ keratitis–ichthyosis–deafness KID syndrome
2728. ✓ keratosis linearis with ichthyosis congenita and sclerosing keratoderma (klick) syndrome
2729. ✓Nephrosis with ichthyosis and adrenal insufficiency
2730. ✓Keratolysis exfoliativa congenita =Continual peeling skin
2731. ✓Generalized inflammatory peeling skin syndrome= Peeling skin syndrome Type B
2732. ✓Generalized peeling skin syndrome= Peeling skin syndrome Type A
2733. ✓Acral peeling skin syndrome
2734. ✓Loricin Keratoderma
2735. ✓Infantile Gaucher disease
2736. ✓Refsum disease=Heredopathis atactica polyneuritiformis
2737. ✓Sjögren-larsson syndrome
2738. ✓Dolichol kinase deficiency
2739. ✓ELOVL4 deficiency syndrome
2740. ✓Tay-sachs syndrome (IBIDS) (congenital ichthyosis with trichothiodystrophy) (Ichthyosis, Brittle hair, Intellectual impairment, Decreased fertility, and Short stature)
2741. ✓Progressive symmetric Erythrokeratoderma= Gottron syndrome= Darier -Gottron syndrome=Erythrokeratodermia figurata variabilis=Symmetrical progressive erythrokeratodermia=Progressive symmetric erythrokeratodermia
2742. ✓Erythrokeratoderma en cocardes
2743. ✓periorificial and ptychotropic erythrokeratoderma PERIOPTER syndrome

2744. ✓Reticular Erythrokeratoderma
2745. ✓Erythrokeratodermia with spinocerebellar ataxia (Giroux–Barbeau syndrome
2746. ✓Erythrokeratodermia-cardiomyopathy syndrome
2747. ✓Erythrokeratoderma variabilis EKV=Mendes da Costa disease=keratosis rubra figurate
2748. ✓Erythrokeratodermia variabilis Cram–Mevorah= Annular erythrokeratodermia figurata variabilis
2749. ✓ Erythrokeratodermia variabilis with erythema gyratum repens
2750. ✓KSDR-related erythrokeratoderma
2751. ✓ Acquired Ichthyosis
2752. Icterus marginatus
2753. Id reaction= autoeczematization=Autosensitization dermatitis
2754. Idiopathic auricular ossification (petrified ear)
2755. Idiopathic dermal melanosis
2756. Idiopathic eruptive hypopigmentation
2757. Idiopathic eruptive macular pigmentation (IEMP)
2758. Idiopathic facial aseptic granuloma (IFAG)
2759. Idiopathic follicular hyperkeratotic spicules
2760. Idiopathic granulomatous mastitis (IGM)
2761. Idiopathic guttate hypomelanosis IGH=leukopathia symmetrica progressiva
2762. Idiopathic lobular panniculitis of pregnancy
2763. Idiopathic lymphoplasmacellular mucositis-dermatitis
2764. Idiopathic mucositis
2765. Idiopathic orbital inflammatory syndrome
2766. idiopathic scrotal calcification
2767. Idiopathic thrombocytopenic purpura
2768. Idiopathic unilateral lower limb gangrene in a neonate
2769. Illusory pigmentation of pseudo-hutchinson's sign
2770. IL-10/IL-10r deficiency
2771. IL-1 receptor-associated kinase-4 (IRAK-4) deficiency
2772. Immersion foot= Trench foot

2773. **Immunoglobulin g4-related disease (IGG4-RD)**
2774. **Immunodeficiency with lymphoid proliferation**
2775. **Immunoglobulin deficiency disorders (see next)**
2776. **✓ Agammaglobulinemia**
2777. **X-linked agammaglobulinaemia (Bruton's disease)**
2778. **✓ Common variable immunodeficiency**
2779. **✓ Selective Ig A immunodeficiency**
2780. **✓ Selective Ig M deficiency**
2781. **✓ Tweak deficiency**
2782. **✓ Activated pi3kδ syndrome**
2783. **✓ x-linked hyper-igm syndrome=Class-switch recombination defects**
2784. **✓ AR hyper-IgM syndromes**
2785. **✓ hypohidrotic ectodermal dysplasia with immunodeficiency**
2786. **✓ immunodeficiency, centromeric instability, and facial anomalies (ICF) syndrome**
2787. **✓ Transient hypogammaglobulinemia of infancy**
2788. **✓ Thymoma with immunodeficiency**
2789. **Impetigo (Non-bullous & Bullous)**
2790. **Impetigo circinate**
2791. **Impetigo of bochart=Superficial pustular folliculitis**
2792. **Impetigo Herpetiformis**
2793. **Impetigo Incognito**
2794. **Impetiginoid Syphiloderm**
2795. **Impetiginization**
2796. **implantation dermatosis (ID)**
2797. **Inclusion body myositis**
2798. **Incidentaloma**
2799. **Indian tick typhus**
2800. **Infantile digital fibromatosis = Inclusion body fibromatosis**
2801. **Infantile gluteal granuloma (papuloerosive dermatitis of Jacquet and Sevestre)**
2802. **Infantile myofibromatosis**

2803. **Infantile pedal papules**
2804. **Infantile perineal (perianal) protrusion**
2805. **Infantile systemic hyalinosis**
2806. **Infantile transient smooth muscle contraction of the skin**
2807. **Infantile tremor syndrome**
2808. **Infective dermatitis associated with HTLV1**
2809. **Inflammatory pseudotumour**
2810. **Inflammatory papillary hyperplasia**
2811. **Incontinentia pigmenti=Bloch–Sulzberger syndrome(inflammatory= vesicular & verrucous & hyperpigmented& hypopigmented=atrophic)**
2812. **Infundibuloma**
2813. **infundibular cyst**
2814. **Injection site reactions**
2815. **Interdigital blastomycetia erosio**
2816. **Interdigital candidiasis**
2817. **Interdigital erythrasma**
2818. **Interdigital gram negative toe web infection**
2819. **Interdigital intertrigo**
2820. **Interdigital pseudomonaus**
2821. **Interdigital tinea pedis**
2822. **Interferon-induced vitiligo**
2823. **Interferon-associated cutaneous necrosis**
2824. **Interstitial granulomatous dermatitis (Ackerman syndrome)**
2825. **Intertrigo**
2826. **Intraoral paratrichosis**
2827. **Intravascular papillary endothelial hyperplasia -Masson's pseudoangiosarcoma**
2828. **Intravascular histiocytosis & intralymphatic histiocytosis (IH)**
2829. **Intraepithelial neoplasia**
2830. **Intrauterine epidermal necrosis**
2831. **Insect bite reaction**
2832. **Invasive fungal dermatitis**
2833. **Inverted follicular keratosis**

Inverse in dermatology (See NEXT)

2834. ✓Acne inversa
2835. ✓Inverse Gottron papules
2836. ✓Inverse halo phenomenon
2837. ✓Inverse klippel-trenaunay syndrome
2838. ✓Inverse koebner phenomeno
2839. ✓Inverse lichen planus
2840. ✓Lichen planus pigmentosus (LPP) inversus
2841. ✓Inverse lichenoid drug eruption
2842. ✓Inverse papular acrokeratosis (acrokeratoelastoides of oswaldo costa)
2843. ✓Inverse pityriasis rosea
2844. ✓Inverse psoriasiform drug eruption
2845. ✓Inverse psoriasis
2846. ✓Inverse sporotrichoid pattern of cutaneous tuberculosis
2847. ✓Inverse subcorneal pustular dermatosis
2848. ✓Inverse tinea versicolor
2849. ✓Inverse type of junctional epidermolysis bullosa (jeb inversa)
2850. ✓Inverse type of recessive dystrophic epidermolysis bullosa (rdeb-i)
2851. ✓Ophiasis inversus
2852. ✓Pterygium inversum unguis (piu)
2853. Invisible dermatoses
2854. Iododerma
2855. IPEX syndrome (immune dysregulation, polyendocrinopathy, enteropathy, x-linked syndrome)
2856. Ipsilateral masseter's spasm
2857. Iron deficiency anemia related skin signs
2858. Ischemia-induced perianal ulceration
2859. Ischemic fasciitis
2860. Isolated melanocytic hyperpigmentation of the distal digits
2861. Isolated occipital white lock
2862. Isolated polymyositis

2863. **Isolated white forelock**
2864. **Isovaleric academia**
2865. **Israeli tick typhus**
2866. **Isthmicoma**
2867. **Isthmus-Catagen cyst**
2868. **Itching purpura= Disseminated pruriginous angiodermatitis**
2869. **Ito hypomelanosis=Incontinentia pigment achromians=systematized achromic nevus**
2870. **Jackson-Sertoli syndrome**
2871. **Jacquet erosive dermatitis**
2872. **Jaffe–Campanacci syndrome**
2873. **Janeway lesions**
2874. **Japanese spotted fever=Oriental spotted fever**
2875. **Johanson–BLizzard syndrome**
2876. **Johnson–Mcmillin syndrome**
2877. **Jogger's nipples=Runner's nipples**
2878. **Joint contractures, muscle atrophy, microcytic anemia, and panniculitis-induced lipodystrophy (jmp) syndrome**
2879. **Juvenile aponeurotic fibroma**
2880. **Juvenile gangrenous vasculitis of the scrotum**
2881. **Juvenile hyaline fibromatosis**
2882. **Juvenile idiopathic arthritis**
2883. **Juvenile melanoma**
2884. **Juvenile temporal arteritis**
2885. **Juvenile plantar dermatosis=Atopic winter feet=forefoot dermatitis =Sweaty sock dermatitis=Dermatitis plantaris sicca**
2886. **Juvenile polyposis syndrome**
2887. **Juvenile Spongiotic Gingivitis**
2888. **Juvenile spring eruption**
2889. **Juvenile xanthogranuloma**
2890. **Kabuki syndrome**
2891. **Kallin syndrome (epidermolysis bullosa simplex with anodontia or hypodontia, nail dystrophy and alopecia)**

2892. **Kanzaki syndrome**
2893. **Kaposi sarcoma (see next)**
2894. **✓Classic**
2895. **✓Bullous**
2896. **✓ Pyogenic granuloma-like kaposi sarcoma (pg-like ks)**
2897. **✓African endemic**
2898. **✓ Iatrogenic**
2899. **✓Immunosuppression related**
2900. **✓AIDS-related epidemic**
2901. **✓pseudo bullous lymphangioma like**
2902. **✓keliodal**
2903. **Kaposi dermatitis= Xeroderma pigmentosum**
2904. **Kaposiform hemangioendothelioma**
2905. **Kaposi-Irgang disease= lupus profundus**
2906. **Kaposi-Stemmer sign**
2907. **Kaposiform lymphangiomatosis**
2908. **Kaposi varicelliform eruption**
2909. **Kasabach-Merritt syndrome=hemangioma with thrombocytopenia**
2910. **Kawasaki disease =Mucocutaneous lymph node syndrome= Kawasaki Syndrom**
2911. **Kayser-Fleischer rings**
2912. **Kearns-Sayre syndrome**
2913. **Keipert syndrome**
2914. **Keloid**
2915. **Ketron-Goodman disease**
2916. **Keratin granuloma**
2917. **Keratoacanthoma= Molluscum sebaceum (See next)**
2918. **✓Solitary**
2919. **✓Giant**
2920. **✓Nodular**
2921. **✓Subungual**
2922. **✓Mucosal**

2923. ✓Centrifugum marginatum
2924. ✓ Generalized eruptive keratoacanthoma of grzybowski
2925. ✓ Multiple keratoacanthomas ferguson-smith syndrome
2926. ✓Chronic involuting relapsing keratoacanthoma
2927. ✓ Multiple familial keratoacanthoma of witten and zak syndrome
2928. Keratocyst= Isthmic-anagen cyst
2929. Keratoderma blennorrhagicum
2930. Keratoderma climatricum =Haxthausen's Disease
2931. Keratoderma simplex
2932. Keratoelastoidosis marginalis of the hands (KEMH)
2933. Keratolysis exfoliativa =Dyshidrosis lamellosa sicca= Recurrent focal palmar peeling
2934. Keratolysis exfoliativa congenita =continual peeling skin
2935. Keratolytic winter erythema=oudtshoorn skin=Erythrokeratolysis hiemalis
2936. Keratosis alba
2937. Keratosis circumscripta
2938. Keratosis follicularis spinulosa decalvans KFSD
2939. Keratosis follicularis squamosa= Dohi's disease
2940. Keratosis lichenoid chronica =Nekam disease=Lichen verrucosus et reticularis=lichenoid trikeratosis=porokeratosis striata lichenoides=lichen ruber moniliformis= keratose lichenoide striae
2941. Keratosis multiformis
2942. Keratosis pilaris= Lichen pilaris
2943. keratosis pilaris atrophicans faciei
2944. Keratosis pilaris decalvans
2945. Keratosis pilaris rubra faceii KPRF
2946. Keratosis rubra pilaris faciei atrophicans
2947. Keratosis palmoplantaris nummularis=hereditary painful callosities
2948. Papular, profuse, precocious keratosis pilaris
2949. Keutel syndrome
2950. Killian syndrome
2951. Kikuchi disease

2952. **Kimura's disease**
2953. **Kindler syndrome =Kindler–Weary syndrome= Bullous acrokeratotic poikiloderma of Kindler and Weary**
2954. **Kirghizian dermato-osteolysis**
2955. **Kirman syndrome**
2956. **Klinefelter syndrome**
2957. **Klippel–Trénauna syndrome=(naevus vasculosus osteohypertrophicus; angio-osteo-hypertrophy syndrome**
2958. **Klippel-Feil syndrome**
2959. **Knuckle pads**
2960. **Kobberling-Dunnigan syndrome**
2961. **Koebner phenomenon**
2962. **Koebner syndrome**
2963. **Koenen tumour**
2964. **Koro syndrome**
2965. **Kosaki overgrowth syndrome**
2966. **Krabbe's disease**
2967. **Kwashiorkor= Protein-calorie malnutrition**
2968. **Kyrle's disease= hyperkeratosis follicularis et parafollicularis in cutem penetrans**
2969. **Labrune syndrome**
2970. **L-2-hydroxyglutaric aciduria**
2971. **Lacrimo-auriculo-dento-digital syndrome LADD**
2972. **Lafora's disease**
2973. **Laddering melanotic pattern of langer's lines (LMPLL)**
2974. **LAMB syndrome (Lentigines, Atrial myxomas, Mucocutaneous myxomas, Blue nevi)**
2975. **Large cell acanthoma**
2976. **Large hematoma occlusion syndrome**
2977. **Larsen syndrome**
2978. **Larsen-like syndrome**
2979. **Larva currens**

2980. **Larva migran =cutaneous larvae migrans (CLM)=Dermatitis verminosa serpiginosa=Creeping Eruption=plumber's itch**
2981. **Laser therapy– induced comedones**
2982. **Lassa fever**
2983. **Late-onset focal dermal elastosis**
2984. **Laugier-Hunziker syndrome**
2985. **Launois-Bensaude syndrome**
2986. **Lawrence syndrome**
2987. **Lawsonella clevelandensis infection**
2988. **Leaflike denture fibroma**
2989. **Legius syndrome**
2990. **Leiomyoma cutis& Piloleiomyomas& Leiomyomas of the Genital Skin**
2991. **Leiomyosarcoma**
2992. **Leigh syndrome**
2993. **Leiner's disease= Erythroderma desquamativum**
2994. **Leishmaniasis (see next)**
2995. **Old World cutaneous leishmaniasis =Baghdad boil= oriental sore (see next)**
2996. **Zoonotic**
2997. **Anthroponotic**
2998. **Leishmaniasis Recidivans=Chronic Lupoid Leishmaniasis**
2999. **Lupoid**
3000. **Dermatomal (Zosteriform) Cutaneous Leishmaniasis**
3001. **Erysipelas - Like Cutaneous Leishmaniasis (Erysipeloid Type)**
3002. **Verrucous Cutaneous Leishmaniasis (Warty CL)**
3003. **Hyperkeratotic Like Cutaneous Leishmaniasis**
3004. **Tumor-Like Cutaneous Leishmaniasis(Pseudo-Tumoral CL)**
3005. **New World cutaneous leishmaniasis= American Leishmaniasis**
3006. **✓ Sarcoid-like**
3007. **✓lupoid**
3008. **✓Ulcerated**
3009. **✓Vegetating**

3010. ✓Erysipeloid
3011. ✓Sporotrichoid
3012. ✓Impetigo-like
3013. ✓verrucous
3014. ✓pustular
3015. ✓plaque-like
3016. ✓Eczematoid
3017. ✓Furunculoid
3018. ✓Miliary
3019. ✓Lichenoid
3020. ✓papulotuberous
3021. ✓Leishmanid
3022. ✓Mucocutaneous
3023. ✓Mucosal leishmaniasis
3024. ✓Disseminated
3025. ✓Diffuse Cutaneous Leishmaniasis= anergic and lepromatous leishmaniasis = pseudolepromatous leishmaniasis= Lepromatoid Leishmaniasis
3026. ✓Visceral= kala-azar=dum-dum fever
3027. ✓post kala-azar Dermal leishmaniasis (PKDL)
3028. Lenaerts syndrome
3029. Inherited pattern Lentigenossis
3030. Acral lentigines (AL)
3031. Centrofacial Lentiginosis
3032. Eruptive lentiginosis
3033. Generalized Lentiginosis
3034. Lentiginosis : oral melanotic macules & Labial melanotic macules & Genital lentiginosis
3035. White Lentiginosis of Grosshans
3036. Lentiginous dysplastic nevus of elderly
3037. Puva- induced lentigines
3038. Segmental lentigenosis= Partial unilateral lentiginosis (PUL)

3039. **Lentigo simplex**
3040. **Solar Lentigo =Senile freckle**
3041. **Ink spot Lentigo**
3042. **Lenz-Majewski syndrome**
3043. **LEOPARD syndrome (Lentigines , ECG abnormalities, Ocular hypertelorism , Pulmonary valve stenosis , Abnormal genitalia, Retardation of growth , and Deafness)**
3044. **Leprechaunism (donohue)**
3045. **Leprosy =Hansen disease=Hanseniasis=Elephantiasis Graecorum (see next)**
3046. ✓ **Borderline leprosy (BT&BL&BB): Borderline Tubercloid & Borderline Lepromatous &Mid borderline**
3047. ✓**Dimorphous leprosy**
3048. ✓ **Histoid leprosy**
3049. ✓**Lazarine leprosy**
3050. ✓ **Indeterminate leprosy**
3051. ✓ **Lepromatous leprosy LL**
3052. ✓**Pure Neuritic Leprosy**
3053. ✓ **Tubercloid leprosy TT**
3054. ✓**Leprosy incognito**
3055. ✓**Lucio leprosy=Diffuse lepromatous leprosy =Pretty leprosy=Lepra bonita**
3056. ✓ **Lucio phenomenon**
3057. ✓**Exacerbation reaction**
3058. ✓**Type 1 reversal reaction T1R**
3059. ✓**Type 2 leprosy reaction= Erythema nodosum leprosum T2R**
3060. ✓**Erythema nodosum Leprosum(necrotican &ulcerative& bullous&Pustular&Target like)**
3061. ✓ **Verrucous leprosy=verrucous leproma**
3062. ✓**Leprosy deformity /deformities/disabilities**
3063. ✓**Rhinomaxillary syndrome of leprosy**
3064. **Leptospirosis**

3065. **Leucinosis**
3066. **Leucocyte adhesion deficiency – LAD (3 types)**
3067. **Leukaemia cutis**
3068. **Leukocytoclastic vasculitis LCV**
3069. **Bullous leukocytoclastic vasculitis (BLCV)**
3070. **Leukodema**
3071. **Leukoderma due to drug**
3072. **Chemical leukoderma**
3073. **Contact leukoderma (contact vitiligo, chemical leukoderma& chemical or contact depigmentation)**
3074. **leukoderma punctata**
3075. **Leukoderma syphiliticum**
3076. **Leukoencephalopathy, brain calcifications, and cysts**
3077. **Leukoplakia**
3078. **Proliferative Verrucous Leukoplakia**
3079. **Oral hairy leukoplakia**
3080. **Leukoplakia with tylosis and esophageal carcinoma**
3081. **Leukotrichia**
3082. **Levamisol =cocaine vasculopathy/levamisole-induced vasculopathy/vasculitis**
3083. **Lichen amyloidosis**
3084. **Lichen aureus= lichen purpuricus**
3085. **Lichen dermatitis**
3086. **Lichen follicularis tumidus**
3087. **Lichen nitidus (generalized & perforating &nail & spinous &linear &purpuric&vesicular &palmoplantar & follicular)**
3088. **Actinic lichen nitidus = summertime actinic lichenoid eruption**
3089. **Lichen nuchae**
3090. **Lichen pilaris**
3091. **Lichen planus LP (see next)**
3092. ✓ **Atrophic**
3093. ✓ **Annular**

3094. ✓ Acute exanthematous
3095. ✓ Hypertrophic
3096. ✓ Bullous Lichen planus
3097. ✓ Lichen planus pemphigoides
3098. ✓ Actinic = Lichen planus actinicus= Lichenoid melanodermatitis= LP tropicus
3099. ✓ Inverse
3100. ✓ Linear
3101. ✓ Zosteriform
3102. ✓vulvovaginal LP
3103. ✓vulvovaginal gingival syndrome
3104. ✓Guttate
3105. ✓ Follicular
3106. ✓Ulcerative LP
3107. ✓ Lichen planus –DLE overlap
3108. ✓oral Lichen planus OLP (Ulcerative & Erosive &Reticulate &Atrophy &Papule &Plaque&Bullous&Pigmented& Mixed)
3109. Lichen planus pigmentosus (Annular&Diffuse& Reticular& Blotchy &Linear&perifollicular& Poikilodermatous)
3110. Lichen planus pigmentosus inversus LPPI
3111. Lichen planopilaris
3112. Lichen Planopilaris Pemphigoides
3113. Lichen ruber acuminatus
3114. Lichen ruber moniliformis
3115. ✓Lichen sclerossus et atrophicus LSEA= Lichen albus
3116. ✓Lichen sclerossus extra genital
3117. ✓Lichen sclerossus bullous hemorrhagic
3118. ✓Hyperkeratotic lichen sclerossus
3119. ✓Lichen sclerossus et atrophicus /morphea overlap
3120. Lichen simplex chronicus LSC
3121. Giant lichenification of Pautrier

3122. **Lichen spinulosus**
3123. **Lichen urticatus**
3124. **Lichen verrucosus et reticularis**
3125. **Lichenoid and granulomatous stomatitis**
3126. **Oral Lichenoid Contact Lesions**
3127. **Oral Lichenoid Drug Reactions**
3128. **Oral lichenoid lesions Related to Graft-Versus- Host Disease (OLL-GvHD)**
3129. **Lichenoid keratosis=lichen planus-like keratoses =Benign lichenoid keratosis**
3130. **Lichenoid granulomatous stomatitis**
3131. **Lichenoid granulomatous dermatitis**
3132. **Lichenoid Dysplasia (LD)**
3133. **Lichenoid pseudovesicular papular eruption**
3134. **Ligature injury**
3135. **Limb girdle muscular dystrophy**
3136. **Limb–Mammary syndrome**
3137. **Linea alba**
3138. **Linear ectodermal cutaneous hamartoma**
3139. **Linear IG A bullous dermatosis LABD= IG A bullous pemphigoid=linear dermatitis herpetiformis=linear IG A dermatosis (lad)**
3140. **Linear naevus syndrome**
3141. **Linea nigra**
3142. **Linea fusca (lf)**
3143. **Linear gingival erythema**
3144. **Linear nevoid hypopigmentation**
3145. **Linear and whorled nevoid hypermelanosis= linear nevoid hyperpigmentation = Zebra-like hyperpigmentation in whorls and streaks**
3146. **Linear focal elastosis = Elastotic striae**

Lines in dermaology (see next)

3147. **✓Antecubital ahn's line**
3148. **✓Aldrich-Mees' lines**
3149. **✓Beau's lines**

3150. ✓Bismuth line
3151. ✓Bunny lines
3152. ✓Blaschko's lines
3153. ✓Burton line=lead line
3154. ✓Dennie–morgan lines/folds
3155. ✓Drug line
3156. ✓Embryonic milk line
3157. ✓Fingerprint lines
3158. ✓Glabellar frown lines
3159. ✓Hart's line
3160. ✓Horizontal necklace lines
3161. ✓Juxtaclavicular beaded lines
3162. ✓kraissl's lines
3163. ✓Marionette lines
3164. ✓Mees' lines
3165. ✓Muehrcke's lines
3166. ✓pastia's lines
3167. ✓pigmentary demarcation lines
3168. ✓Relaxed skin tension lines
3169. ✓Sleep lines
3170. ✓Transverse nasal line
3171. ✓Wallace line
3172. ✓Yellow line of pinkus
3173. ✓Hinderer's lines
3174. Lingual papillitis
3175. Lingual thyroid
3176. Lingual tonsil hypertrophy
3177. Lingual epidermal choristoma
3178. Lip lick dermatitis
3179. Lip pit
3180. Lipedema

3181. **Lipodermatosclerosis =hypodermitis sclerodermaformis= Sclerosing panniculitis=Stasis panniculitis**
3182. **Lipodystrophy**
3183. **✓Lipoatrophia semicircularis**
3184. **✓Annular lipoatrophy**
3185. **✓Congenital generalized lipodystrophy**
3186. **✓Berardinelli-Seip congenital lipodystrophy (BSCL)**
3187. **✓Familial partial lipodystrophy(Dunnigan variant&Kobberling variant)**
3188. **✓Familial partial lipodystrophy associated with mandibuloacral dysplasia**
3189. **✓Acquired generalized lipodystrophy**
3190. **✓Acquired partial lipodystrophy Barraquer–Simons syndrome**
3191. **✓lipodystrophia centrifugalis abdominalis infantilis**
3192. **✓HIV/ART-associated lipodystrophy syndrome**
3193. **✓Recurrent lipoatrophic panniculitis of children**
3194. **✓lipoatrophia annularis (Ferreira-Marques syndrome)**
3195. **✓Localized lipoatrophy**
3196. **✓Localized involutional lipoatrophy**
3197. **✓lipohypertrophy**
3198. **✓Insulin lipodystrophy**
3199. **✓Hemi lipodystrophy**
3200. **Levator ani syndrome**
3201. **Lhermitte-Duclos disease variant of PTEN hamartoma syndrome**
3202. **Lipedematous alopecia (LA)**
3203. **Lipedematous Scalp (LS)**
3204. **Lipogranulomatosis of penis**
3205. **Lipoid proteinosis= hyalinosis cutis et mucosae=Urbach – Wiethe syndrome**
3206. **Lipoma**
3207. **Lipomatosis**
3208. **Lipomatosis of nerve**
3209. **Lipoblastoma**
3210. **Lipoblastomatosis**

3211. **Lipoblastoma-like tumor of the vulva**
3212. **Lipofibromatosis**
3213. **Lipofibromatosis-like neural tumor**
3214. **Lipogranulomatosis**
3215. **Liponeurofibroma**
3216. **Liposarcoma**
3217. **Spindle cell liposarcoma**
3218. **Myxoid liposarcoma**
3219. **Lichtenberg figures**
3220. **Listeriosis**
3221. **Livedo reticularis**
3222. **Livedo racemosa**
3223. **Livedoid vasculopathy = Atrophie blanche**
3224. **Liver diseases &hepatitis &cirrhosis skin signs**
3225. **Lobomycosis= Lobo's disease= keloidal blastomycosis=Lacaziosis**
3226. **Localized hypopigmented keratoses**
3227. **Locus minoris resistentiae**
3228. **Loeffler syndrome**
3229. **Loaiasis= Loa loa filariasis**
3230. **Loeys–Dietz syndrome**
3231. **Löfgren syndrome**
3232. **Loose anagen syndrome**
3233. **Low-grade fibromyxoid sarcoma (LGFMS)**
3234. **Loxoscelism**
3235. **LUBAC (linear ubiquitination assembly complex) components deficiency**
3236. **Lucky winter syndrome**
3237. **Ludwig angina**
3238. **Lues maligna of syphilis**
3239. **Lupus erythematosus (see next)**
3240. **✓Acute cutaneous lupus erythematosus (ACLE) :localized or generalized erythema or bullae,generally associated with SLE**

3241. ✓Subacute lupus (papulosquamous= psoriasiform & annular =lupus marginatus=lupus erythematosus gyratum repens)
3242. ✓Chronic Cutaneous Lupus Erythematosus (CCLE)
3243. ✓Drug-induced lupus erythematosus
3244. ✓Acute syndrome of apoptotic pan-epidermolysis= Toxic epidermal necrolysis-like acute cutaneous lupus erythematosus
3245. ✓ Bullous lupus erythematous
3246. ✓ Chilblain lupus
3247. ✓ DLE discoid lupus erythematosus (localized& disseminated)
3248. ✓Ulcerative DLE
3249. ✓Comedogenic lupus
3250. ✓Pigmented type of chronic cutaneous lupus erythematosus
3251. ✓ Verrucous discoid lupus erythematosus
3252. ✓ Hypertrophic lupus
3253. ✓Lichenoid discoid lupus erythematosus
3254. ✓ Neonatal lupus erythematous
3255. ✓Lupus mastitis LM
3256. ✓Lupus panniculitis
3257. ✓Lupus profundus
3258. ✓Mucosal discoid lupus erythematosus
3259. ✓Lupus erythematosus tumidus
3260. ✓ Systemic lupus erythematous SLE
3261. Lupus miliaris disseminatus facei. LMDF
3262. Lyell's syndrome =Toxic epidermal necrolysis
3263. Lyme disease
3264. Lymphangitis
3265. lymphangitis carcinomatosis
3266. Nodular lymphangitis syndrome
3267. Lymphangiosarcoma= Angiosarcoma
3268. Lymphangioma
3269. Lymphangioma circumscriptum=microcystic lymphatic malformation

3270. **Macrocystic lymphatic malformation=cystic hygroma= lymphangioma cysticum**
3271. **Combined microcystic and macrocystic lymphatic malformation**
3272. **Lymphangiomatosis**
3273. **Lymphangiomyomatosis**
3274. **Kaposiform lymphangiomatosis**
3275. **Lymphangioleiomyomatosis**
3276. **Lymphangiopericytoma**
3277. **Lymphangiectasia**
3278. **Benign Lymphangioendothelioma=Acquired progressive lymphangioma**
3279. **Net-like superficial lymphatic malformation**
3280. **Lymphedema**
3281. **Congenital onset of primary lymphoedema=Milroy disease**
3282. **Primary lymphoedema with later onset=Meige disease**
3283. **Milroy-like disease**
3284. **Hereditary lymphedema**
3285. **Lymphedema-Atrial Septal Defects-Facial Changes syndrome=Irons-Bianchi syndrome**
3286. **Lymphoedema – myelodysplasia =Emberger syndrome**
3287. **Microcephaly with or without chorioretinopathy lymphedema or impaired intellectual development**
3288. **Hennekam lymphangiectasia lymphedema syndrome**
3289. **lymphedema-choanal atresia**
3290. **Hypotrichosis–lymphedema–telangiectasia syndrome**
3291. **Lymphoedema–lipoedema syndrome**
3292. **Massive localized lymphedema**
3293. **Lymphedema praecox**
3294. **Lymphedema tarda**
3295. **Factitial lymphedema**
3296. **Lymphedema–Distichiasis syndrome**
3297. **Lymphedema: filariasis=Elephantiasis tropica (Elephantiasis arabum)**
3298. **Nodular lymphedema**
3299. **Cholestasis – lymphoedema syndrome (Aagenaes syndrome)**

3300. **Generalized, multisegmental lymphatic dysplasia**
3301. **osteopetrosis, lymphoedema, ectodermal dysplasia anhydrotic and immune deficiency syndrome(OLEDAID syndrome)**
3302. **Generalized lymphatic dysplasia (Hennekam syndrome)**
3303. **Microcephaly – lymphoedema – chorioretinal dysplasia**
3304. **Primary lymphedema associated with yellow nails and pleural effusion(yellow nail syndrome)**
3305. **Lymphocele**
3306. **Lymphogranuloma venereum= Lymphogranuloma inguinale**
3307. **Lymphoepithelioma-like carcinoma**
3308. **Adult T-cell leukemia/lymphoma**
3309. **Follicular helper T-cell (TFH) lymphoma**
3310. **Angiocentric T-cell lymphoma**
3311. **Angioimmunoblastic T-cell lymphoma= Angioimmunoblastic lymphadenopathy= Lymphogranulomatosis X**
3312. **CD30-Negative Cutaneous T-Cell Lymphomas**
3313. **CD8-positive pleomorphic small/medium-sized T-cell lymphoma**
3314. **Hydroa vacciniforme–like lymphoma**
3315. **Juvenile granulomatous cutaneous lymphoma (JGCTL)**
3316. **Lymphomatoid papulosis (LYP)**
3317. **Pleomorphic T-cell lymphoma**
3318. **Primary cutaneous acral CD8-positive T-cell lymphoma PCATCL**
3319. **Primary cutaneous anaplastic large cell lymphoma (PC-ALCL)**
3320. **Primary cutaneous CD30-positive lymphoproliferative disorders (LYP)+(PC-ALCL)**
3321. **Primary cutaneous CD4-positive small/medium T-cell lymphoproliferative disorder PCSMTCLPD**
3322. **Primary cutaneous CD8+ aggressive epidermotropic cytotoxic T-cell lymphoma PCAETCL**
3323. **Primary cutaneous gamma/delta γ/δ T-Cell t-cell lymphoma**
3324. **Primary cutaneous peripheral T-cell lymphoma , unspecified**
3325. **Subcutaneous panniculitis-like T-cell lymphoma (SPTCL)**
3326. **Lennert's lymphoma (lymphoepithelioid lymphoma)**

3327. **Lymphoma T cell other ,NOS**
3328. **T cell prolymphocytic leukemia**
3329. **Intravascular T- and NK-cell lymphoma**
3330. **Mycosis fungoides MF (See next)**
3331. ✓ **Hypopigmented Mycosis fungoides**
3332. ✓ **Pigmented purpura –like Mycosis fungoides**
3333. ✓ **Acanthosis nigricans like Mycosis fungoides**
3334. ✓ **Anetodermic Mycosis fungoides**
3335. ✓ **Bullous Mycosis fungoides**
3336. ✓ **Dyshidrosiform mycosis fungoides**
3337. ✓ **Erythema annulare centrifugum like mycosis fungoides**
3338. ✓ **Erythema gyratum repens-like mycosis fungoides (EGR MF)**
3339. ✓ **Folliculotropic mycosis fungoides/ pilotropic**
3340. ✓ **Granuloma annulare like Mycosis fungoides**
3341. ✓ **Granulomatous Mycosis fungoides**
3342. ✓ **Granulomatous slack skin**
3343. ✓ **Hyperpigmented Mycosis fungoides**
3344. ✓ **Ichthyosiform Mycosis fungoides**
3345. ✓ **Interstitial Mycosis fungoides**
3346. ✓ **Invisible mycosis fungoides**
3347. ✓ **keratosis lichenoides like Mycosis fungoides**
3348. ✓ **Poikiloderma vasculare atrophicans=Poikilodermatous mf**
3349. ✓ **Mycosis fungoides associated follicular mucinosis**
3350. ✓ **Mycosis fungoides palmaris et plantaris**
3351. ✓ **Papillomatous/ vegetating**
3352. ✓ **papular mycosis fungoides**
3353. ✓ **perioral dermatitis like mycosis fungoides**
3354. ✓ **pityriasis lichenoides like mycosis fungoides**
3355. ✓ **Psoriasiform mycosis fungoides**
3356. ✓ **pustular Mycosis fungoides**
3357. ✓ **pyoderma gangrenosum like – Mycosis fungoides**

3358. ✓ Syringotropic mycosis fungoides
3359. ✓Adnexotropic (folliculotropic and syringotropic)
3360. ✓ Unilesional/Solitary Mycosis fungoides
3361. ✓ Verrucous/ hyperkeratotic Mycosis fungoides
3362. ✓Erythema chronicum migrans like- t cell lymphoma
3363. ✓Zosteriform mycosis fungoides
3364. ✓ pagetoid reticulosis (ketron-goodman disease& Woringer -kolopp)
3365. Sezary syndrome
3366. Extranodal natural killer (NK)/ T-cell lymphoma (ENKL)
3367. Blastic NK-cell lymphoma=blastic plasmacytoid dendritic cell neoplasm BPDCN= CD4+／CD56+ haematodermic neoplasm
3368. Aggressive NK-cell leukemia (ANKL)
3369. Intravascular T- and NK-cell lymphoma
3370. **B cell Lymphoma (see next)**
3371. ✓ Intravascular diffuse large B-cell lymphoma IVLBCL
3372. ✓ Primary cutaneous diffuse large B cell lymphoma, leg type PCDLBCL
3373. ✓Primary cutaneous follicle centre lymphoma PCFCL
3374. ✓ Cutaneous extranodal marginal zone lymphoma of mucosal-associated lymphoid tissue (MALT)
3375. ✓Primary cutaneous marginal zone lymphoma /lymphoproliferative disorder PCMZLPD
3376. ✓primary cutaneous marginal zone lymphoma (PCMZL)
3377. ✓Secondary cutaneous marginal zone lymphoma (scMZL)
3378. ✓Cutaneous marginal zone lymphomas CMZLs
3379. ✓Anetodermic form of primary cutaneous marginal zone lymphoma
3380. ✓ Primary cutaneous diffuse large B-cell lymphoma, other
3381. ✓B-chronic lymphocytic leukemia/small lymphocytic lymphoma CLL/SLL
3382. EBV-positive mucocutaneous ulcer EBVMCU
3383. EBV-associated diffuse large B-cell lymphoma of the elderly
3384. EBV-positive T- and NK-cell lymphoid proliferations and lymphomas of childhood

3385. **EBV+ diffuse large B-cell lymphoma,not otherwise specified**
3386. **Primary cutaneous mantle cell lymphoma**
3387. **Waldenström's macroglobulinemia/ lymphoplasmacytic lymphoma**
3388. **Precursor T Lymphoblastic leukemia/lymphoma**
3389. **Precursor B lymphoblastic leukemia/lymphoma**
3390. **NK-lymphoblastic leukemia/lymphoma**
3391. **Cutaneous T Lymphoblastic lymphoma**
3392. **Cutaneous B Lymphoblastic lymphoma**
3393. **Lymphomatoid granulomatosis LG**
3394. **Cutaneous amyloidoma**
3395. **Composite lymphomas**
3396. **T-cell/histiocyte-rich B-cell lymphoma**
3397. **Primary effusion lymphoma**
3398. **Cutaneous lymphoid hyperplasia =pseudolymphoma= lymphocytoma cutis= spiegler–fendt sarcoid**
3399. **Predominant T-cell pseudolymphoma**
3400. **Predominant B-cell pseudolymphoma**
3401. **Borrelial lymphocytoma cutis = lymphadenosis benigna cutis**
3402. **Acral pseudolymphomatous angiokeratoma APACHE**
3403. **Pseudolymphoma: miliarial type**
3404. **T-cell rich angiomatoid polypoid pseudolymphoma**
3405. **Drug induced pseudolymphoma**
3406. **Lymphocytic Infiltrate of Jessner= Benign Lymphocytic Infiltrate of the Skin**
3407. **Immunocytoma**
3408. **Extramedullary hematopoiesis**
3409. **Pyogenic lymphoma (neutrophil-rich cd30 anaplastic large cell lymphoma)**
3410. **Autoimmune lymphoproliferative syndrome (Canale–Smith syndrome)**
3411. **EBV-associated lymphoproliferative syndrome**
3412. **Post-transplant lymphoproliferative disorders**
3413. **Methotrexate-associated lymphoproliferative disorder**
3414. **Nodular hyperplasia**
3415. **Lymphoma en cuirasse**
3416. **Plasmacytoma**

3417. **Plasmablastic lymphoma**
3418. **Burkitt lymphoma**
3419. **Hodgkin lymphoma**
3420. **Acute Lymphocytic Leukemia ALL**
3421. **Acute Myeloid Leukemia AML**
3422. **Chronic Lymphocytic Leukemia CLL**
3423. **Chronic Myeloid Leukemia CML**
3424. **T-cell prolymphocytic leukemia (T-PLL)**
3425. **B-cell prolymphocytic leukaemia**
3426. **Hairy cell leukemia**
3427. **pretibial lymphoplasmacytic plaque**
3428. **Lymphoepithelial cyst**
3429. **Lymphoplasmacytic plaque in children(LPC)**
3430. **Lymphorrhoea**
3431. **Lesch -Nyhan syndrome**
3432. **Lynch syndrome**
3433. **Macrocephaly–cutis marmorata telangiectatica congenita**
3434. **MACS (macrocephaly, alopecia, cutis laxa, and scoliosis)**
3435. **Macrodactyly**
3436. **Macrodystrophia lipomatosa**
3437. **Macroglossia**
3438. **Macrophage activation syndrome**
3439. **Macular lymphocytic arteritis=lymphocytic thrombophilic arteritis**
3440. **Maculae cerulae**
3441. **Madelung diseases=Benign symmetric lipomatosis**
3442. **MAGIC syndrome (mouth and genital ulcers with inflammed cartilage)**
3443. **Majeed syndrome**
3444. **Majocchi granuloma=Nodular granulomatous perifolliculitis**
3445. **Maffucci syndrome**
3446. **Malakoplakia = Malacoplakia**
3447. **Malaria**
3448. **Malignant endovascular papillary angioendothelioma =Dabska tumour**
3449. **Malignant endothelial papillary angioendothelioma**

3450. **Malignant fibrous histiocytoma**
3451. **Malignant granular cell tumor**
3452. **Malignant mixed tumor= Malignant chondroid syringoma**
3453. **Malignant triton tumor (MTT)**
3454. **Mabry syndrome**
3455. **Mammary–digital–nail syndrome**
3456. **Mammary-type myofibroblastoma of soft tissue**
3457. **Mammary-type secretory carcinoma of the skin**
3458. **Mandibuloacral macrocephaly, alopecia, cutis laxa and scoliosis (MAC)**
3459. **Mandibuloacral dysplasia**
3460. **Mandibulofacial dysostosis**
3461. **Mannosidosis**
3462. **B-mannosidase deficiency**
3463. **Mantleoma**
3464. **Maple syrup urine disease**
3465. **Marantic endocarditis**
3466. **Marasmus**
3467. **Marfan syndrome**
3468. **Marfanoid progeria–lipodystrophy syndrome**
3469. **Marfanoid hypermobility syndrome**
3470. **Martinez syndrome**
3471. **Marinesco-Sjogren syndrome**
3472. **Marshal –Smith syndrome**
3473. **Marshal White syndrome**
3474. **Mastocytosis (see next)**
3475. ✓ **Adult type mastocytosis**
3476. ✓ **Telangiectasia macularis eruptive perstan TMEP**
3477. ✓ **Mastocytoma**
3478. ✓ **Nodular type**
3479. ✓ **Telangectatic**
3480. ✓ **Diffuse cutaneous mastocytosis**
3481. ✓ **Bullous type**

3482. ✓ Xanthelasmoid= pseudoxanthomatous
3483. ✓ Urticaria pigmentosa=Maculopapular CM
3484. ✓ Mast cell leukemia
3485. ✓ Mast cell sarcoma
3486. ✓ Indolent Systemic mastocytosis
3487. ✓ Aggressive Systemic mastocytosis
3488. ✓ Systemic mastocytosis with associated clonal hematological non-mast cell-lineage disease (sm-ahnmd)
3489. Materia alba
3490. Matrical carcinoma with melanocytic hyperplasia (MCMH) =Malignant melanocytic matricoma
3491. Matting of hair
3492. Maturational hyperpigmentation
3493. Mature plasmacytoid dendritic cell proliferations associated with myeloid neoplasms (MPDMN)
3494. MAUIE syndrome (micropinnae, alopecia universalis, ichthyosis, and ectropion)
3495. MCA/MR Syndrome with Multiple Circumferential Skin Creases
3496. McCune-Albright syndrome
3497. Mcgrath syndrome= Skin fragility syndrome
3498. Mckusick syndrome
3499. MDPL :Mandibular hypoplasia, Deafness, Progeroid features, and Lipodystrophy
3500. Measles=Rubeola= First Disease
3501. Mechanic's hands
3502. Mechanical trauma-induced comedones
3503. Medalion like dendrocyte hamartoma=CD34+ plaque-like dermal fibroma
3504. Median lip fissure
3505. Median raphe cyst
3506. Mediterranean Spotted Fever
3507. Mednik syndrome(mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, keratodermia)

3508. **Megalencephaly capillary malformation MCAP**
3509. **Megalencephaly-polydactyly-polymicrogyria-hydrocephalus (MPPH) syndrome**
3510. **Megarbane–loiselet neonatal progeroid**
3511. **Meige disease**
3512. **Melnick–Fraser syndrome**
3513. **Melkersson–Rosenthal syndrome**
3514. **Mendes da Costa syndrome**
3515. **Melanocytic matricoma**
3516. **Melanosis of the nipple and areola**
3517. **Melanosis Universalis Hereditaria**
3518. **Melanoleucoderma**
3519. **Melanoacanthoma**
3520. **Melanoma (see next)**
3521. ✓ **Acral lentiginous melanoma**
3522. ✓ **Amelanotic melanomas**
3523. ✓ **Balloon cell melanoma**
3524. ✓ **Borderline melanoma& Nevoid melanoma**
3525. ✓ **Malignant blue nevus =blue nevus like melanoma**
3526. ✓ **Clear cell sarcoma=Malignant Melanoma of Soft Tissue**
3527. ✓ **Desmoplastic melanoma**
3528. ✓ **Lentigo maligna = Melanoma in situ**
3529. ✓ **Lentigo maligna melanoma**
3530. ✓ **Melanoma associated hyperpigmentation MAH**
3531. ✓ **Melanoma induced leukoderma**
3532. ✓ **Melanoma metastasis**
3533. ✓ **Blue nevus-like metastatic melanoma**
3534. ✓ **Mucosal melanoma**
3535. ✓ **Nested melanoma of the elderly (NME)**
3536. ✓ **Nodular melanoma**
3537. ✓ **Polypoid nodular melanoma**

3538. ✓ Pigmented epithelioid melanocytomas=Animal type melanoma
3539. ✓Primary dermal melanoma
3540. ✓Small cell melanoma
3541. ✓ Spitzoid melanoma
3542. ✓Subungual melanoma
3543. ✓ Superficial spreading melanoma
3544. ✓ Melanocarcinoma
3545. ✓childhood melanoma
3546. ✓Melanoma in situ
3547. ✓Recurrent melanoma
3548. ✓Verrucous melanoma
3549. ✓MELTUMP (melanocytic tumors of uncertain malignant potential)
3550. ✓Superficial atypical melanocytic proliferation of uncertain significance (SAMPUS)
3551. ✓Intraepidermal atypical melanocytic proliferation of uncertain significance (IAMPUS)
3552. ✓BRCA1-associated protein (BAP1)-inactivated melanocytic tumour (BIMT)
3553. Melanotic neuroectodermal tumor
3554. MELAS: mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes
3555. Melasma =chloasma=Mask of pregnancy
3556. Extra-facial melasma
3557. 308-nm uvb induced melasma
3558. Melioidosis
3559. Melorheostosis
3560. MEND syndrome (male EBP disorder with neurologic defects)
3561. Meningococcemia
3562. Meningomyeloceles
3563. Meningocele
3564. Menkes syndrome

3565. **Mental retardation, enteropathy, deafness, neuropathy, ichthyosis, keratodermia**
3566. **Meralgia paresthetica**
3567. **Merkel cell carcinoma =cutaneous apudoma= primary neuroendocrine carcinoma of the skin**
3568. **Mesonephric cyst**
3569. **Mesothelial cyst**
3570. **Mesotherapy related complications**
3571. **Metageria**
3572. **Metastasis & metastases**
3573. **Methylmalonic acidemia**
3574. **Methylmalonic aciduria**
3575. **3-methylcrotonylcoa carboxylase (ec 6.4.1.4) deficiency**
3576. **Metophyma**
3577. **Mevalonate kinase deficiency**
3578. **Mikulicz syndrome**
3579. **Microcephaly-capillary malformation syndrome**
3580. **Microcephaly-intracranial calcification syndrome**
3581. **Michelin tire baby =Folded skin with scarring =Congenital symmetric circumferential skin creases (CSCSC)**
3582. **Microcystic adnexal carcinoma**
3583. **Microscopic polyangitis= microscopic polyarteritis**
3584. **Microglossia**
3585. **Microvenular hemangioma**
3586. **MIDAS : (Microphthalmia, Dermal Aplasia, and Sclerocornea) =Microphthalmia with linear skin defects(MLS) syndrome**
3587. **Midface toddler excoriation syndrome MiTES**
3588. **Midline anterior neck inclusion cyst**
3589. **Midline skin tag**
3590. **Midline upper lip sinus**
3591. **Mid dermal elastolysis**
3592. **Miescher's radial granulomas**
3593. **Migratory thrombophlebitis**

3594. **Milia & Milium Cysts**
3595. **Milia en plaque**
3596. **Milia like calcinosis**
3597. **Transverse nasal crease milia**
3598. **Milian citrine skin**
3599. **Miliaria=Heat rash=prickly heat**
3600. **Miliaria crystallina**
3601. **Miliaria rubra**
3602. **Miliaria profunda**
3603. **Miliaria pustulosa**
3604. **Miliaria scarlatinosa**
3605. **Miliary haemangiomatosis of infancy**
3606. **Miliary gout**
3607. **Miliary osteoma cutis**
3608. **Miliary tuberculosis**
3609. **Milker nodule= paravaccinia=Pseudocowpox**
3610. **Millipedes**
3611. **Milroy's disease**
3612. **Mitochondrial cytopathies**
3613. **Mitochondrial respiratory chain disorder**
3614. **Mitten-line hyperpigmentation**
3615. **Mixed connective tissue disease(sharp syndrome)**
3616. **Mixed immunobullous disease of childhood**
3617. **Molluscum contagiosum**
3618. **Gianotti-Crosti syndrome-like reaction to molluscum**
3619. **Molluscum-induced comedone**
3620. **Molluscum leprosum**
3621. **Molluscum like histiocytosis**
3622. **Molluscoid Pseudotumor**
3623. **Molluscum fibrosum gravidarum**
3624. **Molluscum pendulum**
3625. **Molluscum sebaceum**
3626. **Mondor's disease= Sclerosing lymphangitis of the penis**

3627. **Mongolian spot =Dermal melanocytosis**
3628. **Monilthrix**
3629. **Monkeypox- Mpox**
3630. **Montgomery tubercle**
3631. **Montgomery syndrome**
3632. **Moraxella infection**
3633. **Morbihan's disease=Solid facial edema**
3634. **MORFAN syndrome (M:mental retardation & O:over growth & RF:remarkable facies & AN:acanthosis negrican)**
3635. **Morgagni-Stewart-Morel syndrome**
3636. **Morgellon disease**
3637. **Morior**
3638. **Morphea =localized scleroderma (see next)**
3639. **✓Plaque-form**
3640. **✓Superficial**
3641. **✓Guttate**
3642. **✓Cordoniform**
3643. **✓ Deep=profunda**
3644. **✓keloidal**
3645. **✓Bullous**
3646. **✓ Linear**
3647. **✓En coup de sabre**
3648. **✓parry-romberg (hemifacial atrophy)**
3649. **✓Generalized**
3650. **✓ Disabling pansclerotic morphea of children**
3651. **✓ Postirradiation morphea**
3652. **Morsicatio buccarum**
3653. **Morsicatio linguarum**
3654. **Morel-lavallee lesion**
3655. **morton neuroma**
3656. **Morvan syndrome**
3657. **Mosaic acral keratosis**

3658. **Mosaic trisomy 8**
3659. **Mosaicism**
3660. **Moynahan syndrome= Xeroderma–talipes–enamel defect**
3661. **Mucinosis (see next)**
3662. **✓Scleromyxoedema =papular mucinosis**
3663. **✓Cutaneous myxoma**
3664. **✓ Cutaneous Focal mucinosis**
3665. **✓Follicular mucinosis /alopecia mucinosa**
3666. **✓Generalized myxedema (myxoedema)**
3667. **✓ Cutaneous Mucinosis of infancy CMI**
3668. **✓Papulonodular mucinosis associated with autoimmune connective tissue diseases**
3669. **✓papular mucinosis of toxic oil syndrome**
3670. **✓Self-healing juvenile cutaneous Mucinosis**
3671. **✓Digital mucous cyst**
3672. **✓Trauma-induced cutaneous mucinosis**
3673. **✓Lichen myxoedematosus**
- **✓Nodular form**
- **✓Discrete papular**
- **✓Acral persistent papular**

3674. **✓ Reticular erythematous mucinosis REM**
3675. **✓ Nevus mucinosus**
3676. **✓Neuropathia mucinosa cutanea**
3677. **✓Nevoid follicular mucinosis**
3678. **✓Obesity associated lymphedematous mucinosis**
3679. **✓Postirradiation localized mucinosis**
3680. **✓Pretibial myxedema (myxoedema)**
3681. **✓Scleredema**
3682. **✓Scleredema of buschke=Scleredema diabeticorum**
3683. **✓Self-healing cutaneous mucinosis**
3684. **✓Urticaria-like follicular mucinosis**

3685. **Mucinous carcinoma**
3686. **Mucinous syringometaplasia**
3687. **Muckle–Wells syndrome**
3688. **Mucocele**
3689. **Mucoepidermoid carcinoma**
3690. **Mucolipidosis**
3691. **Mucopolysaccharidosis**
3692. **✓Hurler-scheie syndrome (mps i-h=s)**
3693. **✓Hurler/Scheie**
3694. **✓ scheie syndrome (mps is)**
3695. **✓ Hunter syndrome (mps ii)**
3696. **✓Sanfilippo syndrome (mps iii):Type A ,B,C,D**
3697. **✓ Morquio syndrome (mp iv):Type A ,B**
3698. **✓ Maroteaux-lamy syndrome (mps vi)**
3699. **✓ Sly syndrome (mps vii)**
3700. **Mucosal neuroma**
3701. **Mucositis**
3702. **Mucormycosis**
3703. **Mudichood**
3704. **Muenke syndrome**
3705. **Muir- Torre syndrome**
3706. **Mukamel syndrome**
3707. **Mulvihill–smith syndrome**
3708. **Mullerian cyst**
3709. **Mumps**
3710. **Multifocal lymphangioendotheliomatosis with thrombocytopenia= Cutaneovisceral angiomatosis with thrombocytopenia**
3711. **Multicentric osteolysis, nodulosis, and arthropathy MONA**
3712. **Multiple cartilaginous exostosis Diaphyseal aclasis**
3713. **Multiple circumscribed lipomas**
3714. **Multiple cutaneous linear neuromas (MCLN)**
3715. **Multiple endocrine neoplasia type MEN 1: Wermer**

3716. **Multiple endocrine neoplasia type 2 (MEN 2A, Sipple syndrome)**
3717. **Multiple endocrine neoplasia MEN type 3 (MEN 2B)= Multiple mucosal neuroma syndrome**
3718. **Multiple hamartoma syndrome**
3719. **Multiple hemangiomas of infancy**
3720. **Multiple minute digitate hyperkeratosis**
3721. **Multiple myeloma**
3722. **Multinucleate cell angiohistiocytoma**
3723. **Multiple pterygia= escobar syndrome**
3724. **Multiple self-healing palmoplantar carcinoma**
3725. **Multiple sulfatase deficiency (mucosulfatidosis)**
3726. **Munchausen syndrome**
3727. **Mycetoma & eumycotic mycetoma & actinomycotic mycetoma & madura foot& maduromycosis**
3728. **Mycoplasma pneumoniae-induced rash and mucositis (MIRM)**
3729. **Myelodysplastic syndrome**
3730. **Myeloid sarcoma**
3731. **Myeloperoxidase deficiency**
3732. **Myeloproliferative disorders :Essential thrombocythemia**
3733. **Myoepithelioma**
3734. **Myoepithelial carcinoma**
3735. **Myopericytoma**
3736. **Myhre syndrome**
3737. **Cutaneous Myiasis(Furuncular Myiasis&Migratory Myiasis (Creeping Myiasis)&Wound Myiasis)**
3738. **Cavitary Myiasis (Nasal& aural&Ocular&Genitourinary)**
3739. **Accidental Myiasis (Pseudomyiasis)**
3740. **Nosocomial Myiasis**
3741. **Myointimoma**
3742. **Adult Myofibromatosis**
3743. **Infantile myofibromatosis=Congenital generalized fibromatosis**
3744. **Myofibroblastic sarcoma=Myofibrosarcoma**

3745. **Inflammatory myofibroblastic tumor =Cutaneous inflammatory pseudotumor**
3746. **Myolipoma**
3747. **Myonecrosis**
3748. **Myotonia**
3749. **Myotonic dystrophy=Steinert disease**
3750. **Myxochondroid metaplasia of the plantar foot**
3751. **Myxofibrosarcoma**
3752. **Myxoinflammatory fibroblastic sarcoma MIFS**
3753. **Myxoid cyst**
3754. **Naegeli-Franceschetti-Jadassohn syndrome**
3755. **Nail dystrophy–Deafness syndrome**
3756. **Nakajo–Nishimura syndrome**
3757. **NAME syndrome (Nevi, Atrial myxoma, Mucinosis of the skin and Ephelides syndrome)**
3758. **Napkin dermatitis (primary irritant contact napkin dermatitis)**
3759. **Allergic contact napkin dermatitis**
3760. **Nasolabial cyst**
3761. **Nasopalatine duct cyst =Cyst of the incisive papilla**
3762. **Natal teeth**
3763. **Natal teeth, patent ductus arteriosus, intestinal pseudo-obstruction**
3764. **Navaratnas in Dermatology(see next)**
3765. **✓Cat eye syndrome**
3766. **✓ Coral bead sign**
3767. **✓Coral dermatitis**
3768. **✓Coral reef bleaching**
3769. **✓Coral red fluorescence**
3770. **✓ Diamond finger gap test**
3771. **✓Diamond hair pattern**
3772. **✓ Diamond peel**
3773. **✓Diamond necklace appearance**
3774. **✓ Diamond's medium**

3775. ✓Diamond keratinocytes
3776. ✓Diamond palmar pattern
3777. ✓ Diamond-shaped depigmented patch (forehead)
3778. ✓ Diamond scales
3779. ✓ Diamond skin disease
3780. ✓ Diamond's triad
3781. ✓ Emerald laser
3782. ✓Epstein pearls
3783. ✓ Gardner-Diamond syndrome
3784. ✓Horn pearls or keratin pearls
3785. ✓ Pearl fractional laser
3786. ✓Pearly penile papules
3787. ✓Pearly wreath
3788. ✓Red coral vessels
3789. ✓ Ruby laser
3790. ✓ Ruby spots
3791. ✓Smegma pearls
3792. ✓String of pearls
3793. ✓Sapphire lasers
3794. Necrobiosis lipoidica
3795. Pigmented necrobiosis lipoidica
3796. Necrobiotic xanthogranuloma
3797. Necrolytic acral erythema NAE
3798. Necrolytic migratory erythema
3799. Necrotizing eosinophilic folliculitis
3800. Necrotizing fasciitis= flesh eating bacteria syndrome
3801. Necrotizing infundibular crystalline folliculitis
3802. Necrotizing lymphocytic folliculitis
3803. Necrotizing neutrophilic dermatosis
3804. Necrotizing sialometaplasia
3805. Neonatal progeroid syndrome=Weidemann-Rautenstrauch

3806. **Neonatal cephalic pustulosis**
3807. **Neonatal cold injury**
3808. **Neonatal genital crisis**
3809. **Neonatal impetigo**
3810. **Neonatal inflammatory skin and bowel disease 1 (NISBD1)**
3811. **Neonatal Onset Multi-Inflammatory disease NOMID**
3812. **Neonatal toxic shock syndrome-like exanthematous disease (NTED)**
3813. **Neonatal varicella**
3814. **Nephrogenic systemic fibrosis**
3815. **Nephrogenic fibrosing dermopathy**
3816. **Néstor–guillermo progeria syndrome**
3817. **NERD: nodules, eosinophilia, rheumatism, dermatitis and swelling**
3818. **Nerve sheath myxoma**
3819. **Peripheral nerve sheath tumor**
3820. **Malignant nerve sheath tumors= Neurosarcoma = Malignant schwannoma**
3821. **Neu laxova syndrome**
3822. **Neural tube defects**
3823. **Neuroblastoma**
3824. **Neurocristic hamartoma**
3825. **Neurocutaneous (leptomeningeal) melanocytosis**
3826. **Neuroendocrine tumours**
3827. **Neurodermatitis**
3828. **Circumscribed neurodermatitis**
3829. **Neurofibroma**
3830. **Plexiform neurofibroma**
3831. **Neurofibromatosis (see next)**
3832. **✓Nf1 von recklinghausen disease**
3833. **✓Nf2 acoustic schwannoma**
3834. **✓Nf3 * mixed (central and peripheral neurofibromas)**
3835. **✓Nf4 * variant**
3836. **✓Nf5 * ,† segmental café-au-lait macules, neurofibromas, or both**
3837. **✓Nf6 * ,‡ café-au-lait macules only**

3838. ✓Nf7 * late onset
3839. ✓Nf-nos not otherwise specified
3840. Neurofibrosarcoma
3841. Neurofollicular hamartoma=spindle cell predominant trichodiscoma
3842. Neurolipomatosis=Neural fibrolipoma= fibrolipomatous hamartoma of nerve
3843. Traumatic Neuroma
3844. Palisaded encapsulated neuroma= Solitary Circumscribed Neuroma
3845. Epithelial sheath neuroma
3846. Neurosarcoma
3847. Neurothekeoma=Dermal nerve sheath myxoma
3848. Atypical cellular neurothekeoma
3849. Neurotic excoriation= Dermatillomania =Skin-picking disorder
3850. Neutral lipid storage disease
3851. ✓Severe congenital neutropenia
3852. ✓Cyclic neutropenia
3853. ✓X-linked neutropenia
3854. ✓Poikiloderma with neutropenia, clericuzio type
3855. Neutrophilic dermatosis of dorsal hands= Pustular Vasculitis
3856. Neutrophilic eccrine hidradenitis NEH
3857. Neutrophilic figurate erythema
3858. Neutrophilic sebaceous adenitis
3859. Neutrophilic superficial eccrine ductitis
3860. Neutrophilic urticarial dermatosis
3861. NEVADA syndrome (nevus epidermicus verrucosus with angiodysplasia and aneurysms)
3862. Nevoid acanthosis nigricans=rounded and velvety epidermal nevus RAVEN
3863. Nevoid basal cell carcinoma syndrome=Gorlin Goltz syndrome= Basal cell nevus syndrome
3864. Nevoid hamartoma
3865. Nevoid hypertrichosis
3866. Nevoid melanoma

3867. **Acquired Nevus (juncional & compound & Intradermal)=Nevocellular Nevus= mole=Cellular Nevus**
3868. **Agminated Nevus**
3869. **Ancient nevus**
3870. **Nevus anelasticus**
3871. **Nevus anemicus- pharmacologic nevus**
3872. **Apocrine nevus**
3873. **Naevus araneus**
3874. **Becker melanosis/nevus=pigmented Hairy epidermal Nevus**
3875. **Becker naevus syndrome**
3876. **Blitz nevus**
3877. **Blue nevus (See next)**
3878. **✓Common**
3879. **✓Cellular:Typical & Atypical**
3880. **✓ Epithelioid**
3881. **✓Malignant Blue nevus =Blue nevus like melanoma**
3882. **✓Target**
3883. **✓Patch like**
3884. **✓Plaque like**
3885. **✓Large plaque-type blue naevus with subcutaneous cellular nodules (LPTBN-SN)**
3886. **✓Hypochromic**
3887. **✓Amelanotic**
3888. **✓Agminate**
3889. **✓Compound**
3890. **✓Combined**
3891. **✓Sclerosing**
3892. **✓Epithelioid and fusiform blue nevus of chronically sun-damaged skin**
3893. **Brauer nevus**
3894. **Cerebriform intradermal nevus CIN**
3895. **Nevus cesius (segmental dermal melanocytosis)**
3896. **Combined Nevus = Biphasic melanocytic nevus**

3897. **Nevus comedonicus**
3898. **Nevus comedonicus syndrome**
3899. **Congenital Nevus (Small & Medium& Large & Giant)**
3900. **Congenital melanocytic nevus (CMN)=Nevus pigmentosus et pilosus=Nevus pilosus**
3901. **Bathing trunk nevus =Giant pigmented nevus**
3902. **Congenital panfollicular nevus**
3903. **Connective tissue nevus**
3904. **Botryoid connective tissue nevus**
3905. **Combined epidermal-connective tissue nevus of proteoglycan**
3906. **Nevus en cocarde /cockade**
3907. **Nevus corniculatus**
3908. **Nevus corymbosa**
3909. **Deep Penetrating nevus**
3910. **Nevus depigmentosa= Achromic nevus**
3911. **Desmoplastic nevus**
3912. **Divided nevus**
3913. **Divided nevus of the eyelid (panda)**
3914. **Divided nevus of the penis**
3915. **Dysplastic nevus=clark nevus= Atypical nevus**
3916. **Eccrine nevus**
3917. **Mucinous Eccrine nevus MEN**
3918. **Eccrine Nevus of the Castori Type**
3919. **Eclipse Nevus**
3920. **Nevus of epidermolysis bullosa**
3921. **Epidermal Nevus EN**
3922. **Epidermolytic Epidermal Nevus**
3923. **Keratinocytic Epidermal Nevus**
3924. **Epidermal Verrucous Nevus**
3925. **Nevus unius lateralis**
3926. **Epidermal naevus – Hailey–Hailey type**
3927. **Epidermal naevus =Darier type = Acantholytic dyskeratotic epidermal nevus**

3928. **Epidermal nevus syndrome= Schimmelpenning–Feuerstein–Mims syndrome**
3929. **Papular epidermal naevus with ‘‘skyline’’ basal cell layer (PENS)**
3930. **Fibroblast growth factor receptor 3 epidermal nevus syndrome (Garcia-Hafner-Happle syndrome)**
3931. **Segmental seborrheic keratosis-like keratinocyte epidermal nevus**
3932. **Hystrix-like epidermal nevus of nevada syndrome**
3933. **Inflammatory linear verrucous epidermal nevus (ILVEN)= Dermatitic epidermal Nevus=Eczematous epidermal Nevus**
3934. **Kerinokeratoticus nevus**
3935. **Glycosaminoglycan Nevi= nevus mucinosus**
3936. **Halo Nevus=Leukoderma acquisitum centrifugum= Sutton’s nevus**
3937. **Hori Nevus(acquired, bilateral nevus of ota- like macules)**
3938. **Nevus of ito=nevus fuscoceruleus acromiodeltoideus**
3939. **Naevus Lentiginosus Linearis**
3940. **Nevus lipomatosis cutaneous superficialis NLCS= Hoffman-Zurhalle nevus**
3941. **Melanocytic nevi of genital skin**
3942. **Melanocytic nevus of acral skin**
3943. **Miescher nevus**
3944. **Meyerson nevus = Eczematous nevi=Halo dermatitis=Halo eczema**
3945. **Naevus marginatus**
3946. **Naevus oligaemicus**
3947. **Oral pigmented nevus**
3948. **Ota nevus =Nevus Fuscoceruleus Ophthalmomaxillaris**
3949. **Sun nevus= nevus of Ota acquisita**
3950. **Nevus osteo nanta**
3951. **Nevus psiloliparus**
3952. **Reactivated nevus**
3953. **Recurrent nevus= pseudomelanoma**
3954. **Rhodoid nevus =capillary malformation-arteriovenous malformation**
3955. **Nevus roseus**
3956. **Sclerosing nevus**
3957. **Nevus sebaceous of Jadassohn**

3958. **Nevus simplex & angel's kiss &stork bite &salmon patch**
3959. **Spark's nevus**
3960. **Nevus spilus=speckled lentiginous nevus=zosteriform lentiginous nevus (Papular Nevus Spilus & Macular Nevus Spilus)**
3961. **Papular Nevus Spilus syndrome**
3962. **Speckled lentiginous nevus syndrome**
3963. **Naevus spinofiliformis**
3964. **✓Spitz Nevus =juvenile melanoma (Agminated& Disseminated)**
3965. **✓Atypical Spitz tumors= STUMP (Spitz Tumour of Uncertain Malignant Potential)**
3966. **✓Malignant =Metastasizing Spitz tumors**
3967. **Pigmented spindle cell nevus of reed**
3968. **Spotted grouped pigmented nevi =Eccrine centered nevus**
3969. **Nevus of striated muscle**
3970. **Nevus sudiferous**
3971. **Targetoid hemosiderotic nevus**
3972. **Nevus tricholemmocycticus**
3973. **Unna nevus = papillated dermal nevus**
3974. **Nevus vascularis mixtus, a hallmark of mixed vascular nevus syndrome (ruggieri- leech syndrome)**
3975. **Nevus vascularis mixtus (cutaneous vascular twin nevi)**
3976. **Large venous nevus**
3977. **Small venous nevi**
3978. **Nevus venous of Servelle- Martorell type**
3979. **Nevus xanthoma**
3980. **Newborn exfoliation= physiological desquamation**
3981. **Newborn and neonatal reactions & diseases**
3982. **Nezelof syndrome**
3983. **Nicolaides–Baraitser syndrome**
3984. **Nicolau syndrome =Embolia cutis medicamentosa**
3985. **Nicotine stomatitis**
3986. **Niemann–pick disease**

3987. **Nijmegen breakage syndrome (NBS)**
3988. **Nipple adenoma**
3989. **Chronic nipple eczema**
3990. **Absent, hypoplastic or anomalous nipples**
3991. **Nevoid hyperkeratosis of nipple**
3992. **Nlrp1-associated autoinflammation with arthritis and dyskeratosis**
3993. **Monogenic nod-like receptor family pyrin domain containing 1 (NLRP1)-associated diseases**
3994. **Nocardiosis**
3995. **Nodular colloid degeneration**
3996. **Nodular elastosis**
3997. **Nodular fasciitis=Nodular pseudosarcomatous fasciitis**
3998. **Nodular scleroderma**
3999. **Nodular vasculitis**
4000. **Nonne-Milroy-Meige syndrome**
4001. **Noonan syndrome**
4002. **Non-suicidal Self-Injury**
4003. **Nontropical diabetic hand syndrome**
4004. **Notalgia parasethica**
4005. **Nuchal-type fibroma**
4006. **Nuchal fibrocartilaginous pseudotumor**
4007. **Nummular dermatitis**
4008. **Nucleotide-binding oligomerization domain (nod)-like receptor family pyrin domain containing 12 (nlrp12)-associated disorder (NLRP12AD)**
4009. **Nuclear factor-kappa b essential modulator (NEMO) deficiency syndrome**
4010. **Oasthouse disease**
4011. **OAS1-associated polymorphic autoinflammatory immunodeficiency disorder (OPAID)**
4012. **Obesity associated lymphedematous mucinosis**
4013. **Obsessive–compulsive disorders**
4014. **Occipital fibrosing alopecia**
4015. **Occipital horn syndrome**
4016. **Ochre Dermatitis**

4017. **Ochronosis**
4018. **Lowe syndrome (Oculo-Cerebro-Renal syndrome)**
4019. **Oculocerebral syndrome with hypopigmentation**
4020. **Oculocerebrocutaneous syndrome (Delleman –Oorthuys syndrome)**
4021. **Oculodental syndrome=Rutherfurd syndrome**
4022. **Oculo-dento-digital dysplasia**
4023. **Oculodermal melanocytosis**
4024. **Oculoectodermal syndrome**
4025. **Oculofaciocardiodental (OFCD)**
4026. **Oculotrichodysplasia**
4027. **Odontogenic cysts**
4028. **Odonto tricho ungual digital palmar syndrome**
4029. **Odonto trichomelic syndrome**
4030. **Odontomicronychial dysplasia**
4031. **Odonto-onycho-dermal ectodermal dysplasia**
4032. **Odonto-onychodysplasia with alopecia**
4033. **Oleoma**
4034. **Oliver–Mcfarlane syndrome**
4035. **Ollier dyschondroplasia**
4036. **Omenn syndrome**
4037. **Omphalomesenteric cyst**
4038. **Omphalomesenteric duct anomalies**
4039. **Omphalocoele**
4040. **Onchocerciasis =river blindness=Erysipelas de la costa=sowda**
4041. **Onychotrichodysplasia and neutropenia,mental retardation**
4042. **Oncocytoma**
4043. **Oral allergy syndrome**
4044. **Oral cancrum oris =Noma**
4045. **Oral facial digital syndrome (See next)**
4046. ✓ **Type i: Papillon-league-psaume syndrome**
4047. ✓**type ii: Mohr syndrome**
4048. ✓**Type iii: Sugarman syndrom**

4049. ✓Type iv: Baraitser-burn syndrome
4050. ✓Type v: Thurston syndrome
4051. ✓ Type vi: Varadi-papp syndrome
4052. ✓ Type vii: Whelan syndrome
4053. ✓ Type viii: oral-facial-digital syndrome, edwards type)
4054. Oral focal mucinosis
4055. Oral melanoma
4056. Oral nevus
4057. Oral papiiloma
4058. Oral Plasma Cell Mucositis
4059. Oral submucous fibrosis
4060. Orange palpebral spots
4061. Orf = Ecthyma contagiosum = Contagious pustular dermatitis
4062. Ornithosis (psittacosis)
4063. Orofacial granulomatosis OFG
4064. Oro-Oculo-Genital syndrome
4065. Osler–Rendu–Weber disease
4066. Osler's nodes
4067. Osteochondroma
4068. Osteochondrolipoma
4069. Osteogenesis imperfect (8 types)
4070. Ossification
4071. Ossifying fibromyxoid tumor OFMT
4072. Ossifying lipoma
4073. Ossifying plexiform tumor
4074. Osteoma cutis
4075. Osteosarcoma
4076. Osteomyelitis
4077. Osteopoikilosis
4078. Oestrogen hypersensitivity vulvovaginitis
4079. Oto-onycho-peroneal syndrome
4080. otopalatodigital syndrome

4081. **Otophyma**
4082. **Otulipenia= Otulin-related autoinflammatory syndrome (ORAS)=otulipenia**
4083. **Oudsthoorn disease**
4084. **p14 deficiency**
4085. **Pachyderma=pachydermia**
4086. **Pachydermatoglyphy**
4087. **Pachydermatous eosinophilic dermatitis**
4088. **Pachydermia oralis**
4089. **Pachydermodactyly**
4090. **Pachydermoperiostosis PDP=Primary Hypertrophic Osteoarthropathy**
4091. **pachyonychia congenita**
4092. **Pachyonychia congenita with amyloidosis and hyperpigmentation**
4093. **Pachyonychia congenita with leukonychia**
4094. **Pacinian corpuscle hyperplasia/pacinian neuroma**
4095. **Paederus/ beetle dermatitis**
4096. **Paget diseases**
4097. **Extramammary paget's disease**
4098. **Pagetoid Dyskeratosis**
4099. **Pagetoid Dyskeratosis of the Scrotum**
4100. **Palatine papilla cyst**
4101. **Palatal exostosis**
4102. **Pallister–killian syndrome**
4103. **Palisaded encapsulated neuroma (PEN)**
4104. **Palisaded neutrophilic and granulomatous dermatitis PNGD**
4105. **Palpable migratory arciform erythema**
4106. **Palmar erythema**
4107. **Palmar fasciitis and polyarthritis syndrome**
4108. **Palmoplantar erythrodysesthesia syndrome**
4109. **Palmoplantar pustulosis**
4110. **Palmoplantar pustular psoriasis**
4111. **Palmoplantar and perioroficial keratoderma with corneal epithelial dysplasia**

4112. **Palmoplantar keratoderma (PPK)**

4113. **Focal palmoplantar keratodermas (ppk)(see next)**

4114. ✓ **Richner–Hanhart syndrome=Oculocutaneous Tyrosinemia**

4115. ✓ **Carvajal Huerta syndrome**

4116. ✓ **Howel–Evans syndrome= Tylosis with esophageal cancer**

4117. ✓ **Pachyonychia congenita type I (Jadassohn–Lewandowsky)**

4118. ✓ **Pachyonychia congenita type II (Jackson–Sertoli)**

4119. ✓ **Pachyonychia congenita type III (Schafer–Branauer)**

4120. ✓ **Pachyonychia congenita type IV (Tarda)**

4121. ✓ **Hereditary keratosis palmoplantaris variant of Wachters**

4122. ✓ **PPK and woolly hair**

4123. ✓ **focal ppk, isolated: Brünauer–Fuhs–Siemens**

4124. ✓ **ppk areata type**

4125. ✓ **striate ppk type 1,type 2,type3**

4126. ✓ **Focal palmoplantar and gingival keratosis**

4127. ✓ **Palmoplantar keratoderma-congenital alopecia 1, Stefanovic type =keratoderma-hypotrichosis-leukonychia totalis syndrome, KHLS)**

4128. ✓ **PPK and congenital alopecia 2, Wallistype (PPKCA2, cataract-alopecia-sclerodactyly syndrome, CASS)**

4129. **Punctate palmoplantar keratodermas (ppk) (see next)**

4130. ✓ **punctate ppk Buschke–Fischer–Brauer (type1)=Keratosis palmoplantaris papulosa**

4131. ✓ **ppk spiny music box= punctate palmoplantar keratoderma type 2 = filiform hyperkeratosis= digitate keratosis**

4132. ✓ **Marginal papular keratoderma: Acrokeratoelastoidosis, Focal acral hyperkeratosis (type 3 punctate ppk)**

4133. ✓ **punctate keratoses of the palmar creases**

4134. ✓ **porokeratosis punctata palmaris et plantaris**

4135. ✓ **Focal acral hyperkeratosis**

4136. ✓ **Papuloverrucous palmoplantar keratoderma of Jakac-Wolf =polykeratosis of touraine**

4137. ✓Cole disease
4138. ✓PLACK syndrome (peeling skin-leukonychia-acral punctate keratosis-cheilitis, and knuckle pad)
4139. **Diffuse palmoplantar keratodermas (ppk)(see next)**
4140. ✓ Nonepidermolytic PPK Unna-Thost
4141. ✓Vörner epidermolytic palmoplantar keratoderma
4142. ✓Unna–Thost epidermolytic palmoplantar keratoderma
4143. ✓Mal de meleda
4144. ✓ Nagashima, kimonis
4145. ✓ Bothnia
4146. ✓Greither epidermolytic palmoplantar keratoderma
4147. ✓Palmoplantar keratoderma of Sybert
4148. ✓Hidrotic ectodermal dysplasia (Clouston) syndrome
4149. ✓KLICK :keratosis linearis, ichthyosis congenita, sclerosing keratoderma
4150. ✓ Bart–pumphrey syndrome=Knuckle pads, leukonychia and deafness
4151. ✓Naxos disease= Mal de Naxos
4152. ✓Schöpf–Schulz–passarge syndrome
4153. ✓Haim–Munk syndromes
4154. ✓ppk mitochondrial
4155. ✓ppk due to connexin 26 defect
4156. ✓Palmoplantar keratoderma with deafness
4157. ✓PPK with nail dystrophy, and motor and sensory neuropathy
4158. ✓Huriez =scleroatrophic and keratotic dermatosis of the limbs
4159. ✓Loricin keratoderma : Camisa variant of Vohwinkel's syndrome
4160. ✓Camisa syndrome
4161. ✓Clouston syndrome
4162. ✓Corneodermatoosseous syndrome
4163. ✓Norbotten
4164. ✓Olmsted syndrome
4165. ✓Papillon–Lefèvre syndrome

4166. ✓Arrhythmogenic right ventricular dysplasia, with mild palmoplantar keratoderma and woolly hair
4167. ✓Diffuse epidermolytic palmoplantar keratoderma (EPPK)
4168. ✓Vohwinkel syndrome =Keratoma hereditarium mutilans
4169. ✓ppk with sex reversal
4170. ✓PPK with pigmentation defects and skin carcinoma
4171. ✓Hyperkeratosis-hyperpigmentation syndrome
4172. ✓Gamborg -Nielsen keratoderma
4173. ✓curly hair–acral keratoderma–caries syndrome
4174. Papillomatoverrucous palmoplantar keratoderma
4175. Keratoderma Associated with Internal Malignancy
4176. Aquagenic transient reactive papulotranslucent acrokeratoderma= Aquagenic syringeal acrokeratoderma
4177. Pangeria
4178. Panfolliculoma
4179. **Panniculitis (see next)**
4180. ✓ Panniculitis due to blunt trauma
4181. ✓ Alpha 1 antitrypsin deficiency panniculitis
4182. ✓ Cold panniculitis
4183. ✓ Cytophagic histiocytic panniculitis
4184. ✓ Factitial
4185. ✓ Pancreatic panniculitis=Enzymic panniculitis
4186. ✓ Post radiotherapy
4187. ✓ Post-steroid
4188. ✓ Rothmann-Makai
4189. ✓ Weber-christian= relapsing febrile nodular non-suppurative panniculitis syndrome
4190. ✓Annular atrophic connective tissue panniculitis of the ankles
4191. ✓Atrophic connective tissue panniculitis
4192. ✓Atypical lymphocytic lobular panniculitis
4193. ✓Crystal deposition panniculitis

4194. ✓Cutaneous polyarteritis nodosa
4195. ✓Equestrian panniculitis
4196. ✓Erythema induratum (nodular vasculitis)
4197. ✓Erythema nodosum
4198. ✓Fasciitis–panniculitis syndrome
4199. ✓Idiopathic lobular panniculitis of pregnancy
4200. ✓Infantile-onset panniculitis with uveitis and systemic granulomatosis
4201. ✓Infection-induced panniculitis
4202. ✓Lipodermatosclerosis
4203. ✓Lipodystrophic panniculitis
4204. ✓Lipophagic panniculitis of childhood
4205. ✓lupus erythematosus panniculitis
4206. ✓Malignancy-related panniculitis-like infiltrates
4207. ✓Morphea/scleroderma panniculitis
4208. ✓Pancreatitis, panniculitis, and polyarthritis (PPP)
4209. ✓Panniculitic bacterid
4210. ✓Panniculitis of dermatomyositis
4211. ✓Panniculitis due to other injectable substances
4212. ✓Panniculitis of lyme disease
4213. ✓Plasma cell panniculitis
4214. ✓Sclerema neonatorum
4215. ✓Subcutaneous fat necrosis of the newborn
4216. ✓Subcutaneous panniculitis -like t -cell lymphoma
4217. ✓Superficial thrombophlebitis
4218. Pantoea (enterobacter) agglomerans
4219. PAC (Pyoderma gangrenosum, Acne and Ulcerative colitis)
4220. PAPA syndrome (Pyogenic arthritis, pyoderma gangrenosum and Acne)
4221. PAPASH syndrome (Pyogenic arthritis, Acne, Pyoderma gangrenosum and Suppurative hidradenitis)
4222. PASH syndrome (Pyoderma gangrenosum , Acne and Suppurative hidradenitis)

4223. **PASS (Pyoderma gangrenosum, Acne, and Ankylosing spondylitis, with or without Suppurative hidradenitis)**
4224. **PSAPASH (psoriatic arthritis, pyoderma gangrenosum, Acne and Hidradenitis suppurativa)**
4225. **Papillary carcinoma**
4226. **Papillary eccrine adenoma=Tubular apocrine adenoma**
4227. **Papillary endothelial hyperplasia=Masson's tumor**
4228. **Papillary hemangioma**
4229. **Papillary hyperplasia of the palate**
4230. **Papillary intralymphatic angioendothelioma (PILA)=Dabska-type hemangioendothelioma=dabska tumor=endovascular papillary angioendothelioma**
4231. **Papillomatosis cutis carcinoides Gottron**
4232. **Papillomatosis cutis lymphostatica**
4233. **Papillon–Léage syndrome**
4234. **Papillon–Léfèvre syndrome**
4235. **Papular Acantholytic Dyskeratosis**
4236. **Papular clear cell hyperplasia of the eccrine duct**
4237. **Papular dermatitis= chronic papular urticarial= Chronic prurigo of adults**
4238. **Papular epidermal nevus with skyline**
4239. **Papular eruption of blacks**
4240. **Papular purpuric gloves and socks syndrome**
4241. **Papuloerythroderma of Ofuji**
4242. **Parachordoma**
4243. **Paracoccidioidomycosis=South american blastomycosis**
4244. **Paraffinoma**
4245. **Paraganglioma-like dermal melanocytic tumor**
4246. **Paraquat tongue**
4247. **Paragonimiasis**
4248. **Parapox**
4249. **Parakeratosis variegate**
4250. **parameatal urethral cyst**
4251. **Paraneoplastic erythroderma**

4252. **Paraneoplastic scleroderma-like syndrome**
4253. **Paraneoplastic pemphigus PNP**
4254. **Paraneoplastic autoimmune multiorgan syndrome PAMS**
4255. **Small plaque Parapsoriasis =chronic superficial scaly dermatitis (Digitate dermatosis & Xanthoerythrodermia perstans)**
4256. **Large plaque Parapsoriasis (parapsoriasis variegate =parakeratosis variegata =Retiform parapsoriasis= parapsoriasis lichenoides & poikilodermatous Parapsoriasis)**
4257. **Parasitophobia**
4258. **Parinaud syndrome**
4259. **Parkes Weber syndrome**
4260. **Paroxysmal nocturnal hemoglobinuria**
4261. **Parry-Romberg syndrome**
4262. **Parulis**
4263. **Pasteurellosis**
4264. **pearly penile papules**
4265. **Pebbling**
4266. **PEComa (Fibroma-like perivascular epithelioid cell tumor)**
4267. **pediculosis capitis (Head lice)**
4268. **Pediculosis corporis**
4269. **Pediculosis pubis & pthirus pubis=crab lice**
4270. **Phthiriasis palpebrarum**
4271. **Pediculid**
4272. **peeling skin syndromes**
4273. **Pellagra**
4274. **Perlman syndrome**
4275. **Pemphigus erythematous = Senear-Usher syndrome**
4276. **Erythema annulare centrifugum-like pemphigus erythematosus (EAC-like PE)**
4277. **Pemphigoid gestationis =herpes gestation=Gestational pemphigoid**
4278. **Pemphigus foliaceus**
4279. **Endemic pemphigus foliaceus= Fogo selvage= Brazilianpemphigus foliaceus**
4280. **Pemphigus herpetiformis**

4281. **IgA Pemphigus :Intraepidermal neutrophilic type**
4282. **IgA Pemphigus : Subcorneal pustular dermatosis**
4283. **Pemphigus syphiliticus**
4284. **Contact pemphigus**
4285. **Pemphigus vegetans(Hallopeau type & Neumann type)**
4286. **Pemphigus vulgaris**
4287. **Juvenile pemphigus vulgaris**
4288. **Verrucous pemphigus=pemphigus acanthomata=postpemphigus acanthoma**
4289. **Neonatal pemphigus vulgaris**
4290. **Pena-shokeir syndrome**
4291. **Pendulous fibroma**
4292. **Pencil-core granuloma**
4293. **Penicillamine-induced elastosis perforans**
4294. **Penicilliosis**
4295. **Penile acne**
4296. **Carcinoma of the penis**
4297. **Penile intraepithelial neoplasia**
4298. **Penile lymphedema**
4299. **Penile phimosis &paraphimosis**
4300. **Penile plasma cell (zoon's) balanitis**
4301. **Penile sclerosing lymphangitis**
4302. **Penttinen syndrome**
4303. **Perianal and vulvovaginal (perineal) streptococcal infection**
4304. **Perianal comedones**
4305. **Perianal streptococcal dermatitis**
4306. **Periarticular thenar erythema with onycholysis (PATEO)**
4307. **Periflexural exanthema**
4308. **Perifollicular elastolysis**
4309. **Perifollicular fibroma**
4310. **Perinatal gangrene of the buttock**
4311. **Recurrent toxin-mediated (perineal) erythema**

4312. **Perineurioma=storiform perineurial fibroma=localized hypertrophic neuropathy=Storiform perineurial fibroma=Fibrolamellar nerve sheath tumor**
4313. **Malignant perineurioma**
4314. **Sclerosing perineurioma**
4315. **Periodontitis**
4316. **Periodontal fistula**
4317. **Periodic fever with aphtous stomatitis, pharyngitis and adenitis syndrome**
4318. **Periodic fever, immunodeficiency, and thrombocytopenia**
4319. **Perioral dermatitis**
4320. **Periobital acquired dermal macular hyperpigmentation**
4321. **Periorificial dermatitis=childhood granulomatous periorificial dermatitis**
4322. **Peristomal skin related skin disorders**
4323. **Periumbilical calcific elastolysis**
4324. **Periumbilical perforating pseudoxanthoma elasticum**
4325. **Peroxisomopathies**
4326. **Peripheral giant cell granuloma**
4327. **Peripheral odontogenic fibroma**
4328. **Peripheral ossifying fibroma**
4329. **Periporitis staphylogenes = Sweat gland abcess**
4330. **Pernio = Chilblains**
4331. **Perniola syndrome**
4332. **Perlèche**
4333. **Petechiae**
4334. **Persistent cholinergic erythema**
4335. **Persistent acrovasculopathy syndrome**
4336. **Persistent Linear Bands in Infancy**
4337. **Peutz-Jeghers syndrome**
4338. **Peyronie's disease**
4339. **Peyronellaea gardeniae infection**
4340. **PFAPA :periodic fever, aphthae, pharyngitis, and Adenitis syndrome**
4341. **Phalangeal sarcoma**

4342. **Phacomatosis & phakomatosis (see next)**
4343. ✓ **Type i (Adamson-Best syndrome)**
4344. ✓ **Type ii (Takano-Kruger-Doi syndrome)**
4345. ✓ **Type iii (Kobori-Toda syndrome)**
4346. ✓ **Type iv**
4347. ✓ **phakomatosis cesioflammea**
4348. ✓ **phakomatosis spilorosea**
4349. ✓ **phakomatosis melanovascularis**
4350. ✓ **phakomatosis cesiomarmorata**
4351. ✓ **phakomatosis pigmentovascularis**
4352. ✓ **phakomatosis pigmentokeratotica**
4353. **Phakomatosis Pigmento-Eccrina**

Phenomena in dermatology (see next)

4354. ✓ **Alopecia areata (aa) sparing lesional gray/white hairs**
4355. ✓ **Anatopic phenomenon**
4356. ✓ **Atopic dermatitis generally spare the napkin area**
4357. ✓ **Bell's phenomenon**
4358. ✓ **Bork-Baykal phenomenon**
4359. ✓ **Brocq's phenomenon**
4360. ✓ **Butterfly sign**
4361. ✓ **Castling phenomenon**
4362. ✓ **Deck chair sign**
4363. ✓ **Delayed blanch phenomenon**
4364. ✓ **Ear lobe sign**
4365. ✓ **Epidermal growth factor receptor (egfr) inhibitor-associated rash spares irradiated areas**
4366. ✓ **Epitope spreading phenomenon**
4367. ✓ **Halo phenomenon**
4368. ✓ **Harlequin phenomenon**
4369. ✓ **Hoigne's phenomenon**
4370. ✓ **ichthyosis vulgaris spares flexures**

4371. ✓ irritant diaper dermatitis spares flexures
4372. ✓ islands of sparing in pityriasis rubra pilaris
4373. ✓ Isocaumatopic nonreaction
4374. ✓ Isocaumatopic reaction
4375. ✓ Isodermatopic nonreaction
4376. ✓ Isodermatopic reaction
4377. ✓ Isolymphtopic nonreaction
4378. ✓ Isolymphtopic reaction
4379. ✓ Isomosaictopic nonreaction
4380. ✓ Isomosaictopic reaction
4381. ✓ Isoneuraltopic nonreaction
4382. ✓ Isoneuraltopic reaction
4383. ✓ Isophototopic nonreaction
4384. ✓ Isophototopic reaction
4385. ✓ Isoradiotopic nonreaction
4386. ✓ Isoradiotopic reaction
4387. ✓ Isoscartopic nonreaction
4388. ✓ Isoscartopic reaction
4389. ✓ Isoshockwave nonreaction
4390. ✓ Isoshockwave reaction
4391. ✓ Isotattotopic nonreaction
4392. ✓ Isotattotopic reaction
4393. ✓ Isotraumatopic nonreaction
4394. ✓ Isotraumatopic reaction
4395. ✓ Isovaccinetopic nonreaction
4396. ✓ Isovaccinetopic reaction
4397. ✓ Isovenostatic nonreaction
4398. ✓ Isovenostatic reaction
4399. ✓ Kasabach merritt phenomenon
4400. ✓ knuckle sparing of acute cutaneous lupus erythematosus (ACLE)
4401. ✓ koebner reaction

4402. ✓ **Lucio phenomenon**
4403. ✓ **Mauserung phenomenon**
4404. ✓ **Meirowsky phenomenon**
4405. ✓ **Meyerson phenomenon**
4406. ✓ **Milian's ear sign**
4407. ✓ **Nasolabial fold sparing in the malar rash of systemic lupus erythematosus**
4408. ✓ **Nikolskiy's phenomenon**
4409. ✓ **Nose sign**
4410. ✓ **Nottus phenomenon**
4411. ✓ **Olfleck's phenomenon**
4412. ✓ **Pagetoid phenomenon**
4413. ✓ **paretic limbs spared in rheumatic diseases**
4414. ✓ **Pathergy phenomenon**
4415. ✓ **pityriasis rosea spare sun-exposed areas**
4416. ✓ **Prozone phenomenon**
4417. ✓ **pruritic urticarial papules and plaques of pregnancy (puppp) spares periumbilical area**
4418. ✓ **psoriasis often spare central face**
4419. ✓ **Raynaud's phenomenon**
4420. ✓ **Rebound phenomenon**
4421. ✓ **Relapsing polychondritis spare earlobes**
4422. ✓ **Renbok phenomenon**
4423. ✓ **Reverse koebner phenomenon**
4424. ✓ **Reverse koebner reaction**
4425. ✓ **Reynold's braude phenomenon**
4426. ✓ **Rosacea spares circum-oral and peri-orbital areas**
4427. ✓ **Rumpel leede phenomenon**
4428. ✓ **scrotum sparing in tinea cruris**
4429. ✓ **spared zones in leprosy**
4430. ✓ **Splendore-hoeppli phenomenon**

4431. ✓ **Sulzberger-chase phenomenon**
4432. ✓ **sunbed suntan sacroscapular sparing sign**
4433. ✓ **Ultraviolet recall phenomenon**
4434. ✓ **upper axillary area spared in pityriasis versicolor**
4435. ✓ **vermilion border spared in an acrodermatitis enteropathica (ae)-like syndrome**
4436. ✓ **vermillion border spared in peri-oral dermatitis**
4437. ✓ **white islands in a sea of red in dengue fever**
4438. ✓ **wilkinson's triangle**
4439. ✓ **wolf 's isotopic response**
4440. ✓ **wolf's isotopic nonreaction**
4441. **Phenylketonuria PKU**
4442. **Phaeohyphomycosis**
4443. **Phlebolymphedema**
4444. **Phlebectasia**
4445. **Phosphaturic mesenchymal tumor**
4446. **Photoageing &Photoaging**
4447. **Photodamage**
4448. **Photodistributed eruptive telangiectasia**
4449. **Photoinduced acute exanthematous pustulosis**
4450. **photolocalized purpura**
4451. **Photosensitive spongiotic/lichenoid eruption of micropapules and plaques**
4452. **Phototoxic reaction**
4453. **Photoallergic reaction**
4454. **Photoleukomelanodermatitis of kobori**
4455. **Photo-onycholysis**
4456. **Phrynoderma**
4457. **Phylloid hypomelanosis**
4458. **Phylloid hypermelanosis**
4459. **Phylloid terminal hair nevus**
4460. **Physical purpura**
4461. **Physiological desquamation**

4462. **Physiologic hyperpigmentation**
4463. **Physiological melanoplakia**
4464. **Phytophotodermatitis**
4465. **PIBIDS (photosensitivity, Brittle nail , ichthyosis, brittle hair, intellectual impairment, decreased fertility and short stature)**
4466. **PBIDS (Photosensitivity-Brittle hair-Brittle nail-intellectual impairment-Decreased fertility-Short stature syndrome)**
4467. **Picker's nodule**
4468. **Piebaldism = white spot disease**
4469. **Black Piedra = Trichomycosis nodularis**
4470. **White piedra= Trichosporosis nodosa**
4471. **Pierre robin syndrome**
4472. **Piezogenic pedal papules**
4473. **Piezogenic wrist papules**
4474. **pigmented allergic salute sign**
4475. **Pigmented follicular cyst**
4476. **Pigmentary damarcation lines PDL= Futcher's lines=Voigt lines= Ito's lines**
4477. **Pigmentary mosaicism**
4478. **pigmentary twin spotting**
4479. **Pigmented macules of bony prominences (PMBP)**
4480. **Pigmented neuroectodermal tumour of infancy**
4481. **Pigmented purpuric dermatosis PPD (see next)=purpura pigmentosa chronica**
4482. ✓ **Eczematid-like purpura of Doucas and kapetanakis**
4483. ✓ **Lichen aureus=lichen purpuricus**
4484. ✓ **Familial pigmented purpuric eruption**
4485. ✓ **pigmented purpuric lichenoid dermatitis of Gougerot and Blum=lichenoid purpura = purpuric pigmented lichenoid dermatitis**
4486. ✓ **Linear pigmented purpura**
4487. ✓ **Granulomatous pigmented purpura**
4488. ✓ **Purpura annularis telangiectodes of Majocchi**
4489. ✓ **Schamberg disease =progressive pigmentary dermatosis**

4490. **pigmented transverse nasal band**
4491. **Pigmentatio reticularis faciei et colli**
4492. **Pik3ca-related segmental overgrowth syndrome PROS**
4493. **Pilar cyst= wen =Trichilemmal cyst= Isthmus-catagen cyst**
4494. **Pilar sheath acanthoma**
4495. **Proliferating pilar tumor**
4496. **Proliferating trichilemmal cyst**
4497. **Malignant proliferating trichilemmal tumors**
4498. **Pili annulati**
4499. **Pseudopili annulati**
4500. **Pili bifuracti**
4501. **Cutaneous pili migrans (CMP)**
4502. **Pili multigemini**
4503. **Pili torti**
4504. **Pili torti and developmental delay**
4505. **Pili torti and onychodysplasia (Beare type)**
4506. **Pili torti and onychodysplasia**
4507. **Pili Canaliculi et Trianguli =Uncombable hair syndrome=Spun glass hair**
4508. **Pilodental dysplasia with refractive errors**
4509. **Pilomatricoma/ Pilomatrixoma= calcifying epithelioma of Malherbe**
4510. **Pilomatricoma variants (Anetodermic &Bullous &Exophytic&Familial &Giant& Lymphangiectatic &Multiple&Perforating & Proliferating & Pseudo bullous&Ulcerative)**
4511. **Pilomatricomal horn**
4512. **Pilomatrical carcinoma**
4513. **Pilomatrical carcinosarcoma**
4514. **Pilomatrixcarcinoma**
4515. **Pilonidal sinus**
4516. **Pinta**
4517. **Pintoid dyschromia of yaws**
4518. **Pipkin syndrome**

4519. **Pityriasis rubra pilaris PRP=Lichen ruber pilaris = Lichen Ruber acuminatus**
- ✓ **Classic adult**
- ✓ **Atypical adult**
- ✓ **Classic juvenile**
- ✓ **Circumscribed juvenile**
- ✓ **Atypical juvenile**
- ✓ **Human immunodeficiency virus infection associated**

4520. **Pityriasis alba= Impetigo pityroides= Pityriasis simplex= Pityriasis maculata**
4521. **Pityriasis folliculorum**
4522. **Pityriasis lichenoid chronica PLC**
4523. **Pityriasis lichenoides et varioliformis acuta PLEVA**
4524. **Febrile ulceronecrotic variant of PLEVA =Febrile Ulceronecrotic Mucha-Habermann disease FUHMD =Pityriasis lichenoides with Ulceronecrosis and Hyperthermia**
4525. **Pityriasis rosea (Classic & Vesicular & Urticarial & Target like& Purpuric & Pustular & Abortive&Inverted&Localized&Segmental&Unilateral)**
4526. **pityriasis rosea Gibert**
4527. **Pityriasis rosea gigantea of Darier**
4528. **Pityriasis circinata et marginata of vidal**
4529. **Pityriasis rotunda= Pityriasis Circinata**
4530. **Pityriasis Steatoides**
4531. **Pitted keratolysis = keratolysis plantare sulcatum**
4532. **Pfeiffer syndrome**
4533. **Plague**
4534. **PLAID immune plcg2-associated antibody deficiency and immune dysregulation**
4535. **APLAID (autoinflammation and plcg2-associated antibody deficiency and immune dysregulation)**
4536. **Plantar fibromatosis = Ledderhose disease**
4537. **Plaque-like CD34-positive dermal fibroma**
4538. **Plaque-like myofibroblastic tumor**

4539. **Plasma cell gingivitis =Allergic gingivostomatitis= Idiopathic gingivostomatitis= Atypical gingivostomatitis**
4540. **Plasma cell glossitis**
4541. **Plasma cell granuloma**
4542. **Plasmacytoma**
4543. **Plasmacytosis**
4544. **Plasmoacanthoma**
4545. **Plate-like osteoma cutis**
4546. **Pleomorphic adenoma**
4547. **Carcinoma ex pleomorphic adenoma**
4548. **Pleomorphic dermal sarcoma**
4549. **Pleomorphic fibroma of the skin**
4550. **pleomorphic hyalinizing angiectatic tumor**
4551. **Pleomorphic lipoma**
4552. **Pleomorphic liposarcoma**
4553. **Pleomorphic Sarcoma**
4554. **Undifferentiated pleomorphic sarcoma**
4555. **Pleonosteosis**
4556. **Plexiform fibrohistiocytic tumor**
4557. **Plica polonica=Plica neuropathica**
4558. **Plummer-vinson syndrome**
4559. **Pneumocystis carinii=pneumocystosis**
4560. **Podoconiosis**
4561. **POEMS syndrome (polyneuropathy, organomegaly, endocrinopathy, M-protein and skin changes)= crow-fukase syndrome**
4562. **Pohl–Pinkus constriction**
4563. **Poikiloderma**
4564. **Poikiloderma vasculare atrophicans**
4565. **Poikiloderma-like cutaneous amyloidosis**
4566. **Poikiloderma of civatte**
4567. **Poikilodermatous plaque-like hemangioma (PPLH)**
4568. **Pointlist melanotic macules**
4569. **Poison ivy dermatitis**

4570. **Poland syndrome**
4571. **Poliosis**
4572. **Pomade crust**
4573. **polyarteritis nodosa =panarteritis nodosa PAN**
4574. **Polycythemia rubra vera=Erythremia**
4575. **Polycystic ovarian syndrome (PCOS)**
4576. **Polymastia**
4577. **Polymorphous cold eruption**
4578. **Polymorphous light eruption PMLE**
4579. **polymorphous sweat gland carcinoma**
4580. **Polymorphous (low-grade) adenocarcinoma**
4581. **Polymyositis PM(PM alone &PM associated with malignancy& as a part of an overlap connective tissue disorder)**
4582. **Polyostotic fibrous dysplasia**
4583. **Polythelia pilosa**
4584. **Pompholyx**
4585. **pool palms/pool toes**
4586. **Popliteal pterygium syndrome= klein syndrome**
4587. **Popsicle panniculitis**
4588. **Porokeratosis (see next)**
4589. **✓Porokeratosis of Mibelli**
4590. **✓Disseminated superficial actinic porokeratosis (DSAP)**
4591. **✓ linear porokeratosis**
4592. **✓punctate porokeratosis**
4593. **✓Eruptive disseminated porokeratosis**
4594. **✓Follicular porokeratosis (FPK)**
4595. **✓Bullous Porokeratosis**
4596. **✓Porokeratosis palmaris et plantaris et disseminata PPPD**
4597. **✓Eruptive pruritic papular porokeratosis**
4598. **✓porokeratosis ptychotropica**
4599. **✓Postirradiation multiple minute digitate porokeratosis**
4600. **✓Verrucous porokeratosis**

4601. ✓porokeratotic eccrine ostial and dermal duct nevus (PEODDN)
4602. ✓Porokeratotic eccrine and hair follicle nevus PEHFN
4603. ✓Porokeratotic Adnexal Ostial Nevus (PAON) = PEHFN and PEODDN
4604. ✓Porokeratoma=porokeratotic acanthoma
4605. Linear palmo-plantar porokeratotic hamartoma
4606. Poroma
4607. Porocarcinoma
4608. **Porphyria (see next)**
4609. ✓ ALA dehydratase deficiency
4610. ✓ Acute intermittent porphyria
4611. ✓ Congenital erythropoietic porphyria= gunther syndrome CEP
4612. ✓ Hereditary coproporphyria
4613. ✓ Porphyria cutaneous tarda PCT
4614. ✓ Erythropoietic protoporphyria EPP
4615. ✓ Hepatoerythropoietic porphyria
4616. ✓ Pseudoporphyria= PCT-like syndrome=bullous dermatosis of hemodialysis=drug-induced pseudoporphyria cutanea tarda
4617. ✓ x-linked dominant protoporphyria
4618. ✓ Porphyria variegate=South African porphyria
4619. Portwine stain=nevus flammeus
4620. Postcardiotomy syndrome
4621. Post ECG leads dermatitis
4622. Post Herpetic neuralgia
4623. Post-implantation erythema (PIE)
4624. Post inflammatory hyperpigmentation/postinflammatory melanosis
4625. Post inflammatory hypopigmentation/postinflammatory leukoderma
4626. Postirradiation digitate keratosis (PIDK)
4627. Post-Irradiation pseudo-sclerodermatous panniculitis
4628. Postmastectomy Lymphedema
4629. Neutrophilic Dermatosis on Postmastectomy Lymphedema
4630. Post Menopausal symptoms
4631. Post-Miliarial exfoliation= post-heat rash exfoliation

4632. **Post-Stripping cutaneous sclerosis**
4633. **Post-Thermal burn pruritus**
4634. **Post-Traumatic erosive dermatosis of the scalp**
4635. **Post Traumatic hematoma**
4636. **Pott's puffy tumor**
4637. **Powassan virus related skin infection**
4638. **Prader willi syndrome**
4639. **Prayer nodule**
4640. **preauricular sinus**
4641. **Precalcaneal congenital fibrolipomatous hamartoma**
4642. **Pre- and postnatal growth retardation, mental retardation, and acral limb deficiencies**
4643. **Prepubertal Periodontitis**
4644. **Prepubertal unilateral fibrous hyperplasia of the labium majus**
4645. **Prepubertal vulval fibroma=childhood asymmetric labium majus enlargement**

Pregnancy diseases (see next)

4646. **✓Atopic eruption of pregnancy = prurigo of pregnancy=Prurigo gestationis (besnier) =Early-onset prurigo of pregnancy = Papular dermatitis of pregnancy = Pruritic folliculitis of pregnancy = Linear igm disease of pregnancy = Eczema in pregnancy**
4647. **✓Impetigo herpetiformis= Pustular psoriasis of pregnancy**
4648. **✓Pupp (pruritic urticarial papules and plaques of pregnancy= Toxic erythema of pregnancy=Late-onset prurigo of pregnancy = Polymorphic eruption of pregnancy = Toxemic rash of pregnancy**
4649. **✓Intrahepatic cholestasis of pregnancy = Obstetric Cholestasis**
4650. **✓prurigo gravidarum**
4651. **✓Pregnancy-Related Pyogenic Granuloma (Granuloma Gravidarum)**
4652. **✓Other physiologic changes**
4653. **premature gray/grey hair**
4654. **Pressure purpura (cough &vomiting)**
4655. **Pretibial angioplasia**

4656. **Primary signet-ring cell/histiocytoid carcinoma**
4657. **Primary cutaneous adenoid cystic carcinoma**
4658. **Primary cutaneous adenomyoepithelioma (pcAME)**
4659. **Primary cutaneous angioplasmacellular hyperplasia**
4660. **Primary cutaneous apocrine carcinomas / adenocarcinoma**
4661. **primary cutaneous mucinous carcinoma (PCMC)**
4662. **Primary cutaneous cribriform carcinoma (PCCC)**
4663. **Primary cutaneous extraskeletal osteosarcoma**
4664. **Primary lymphoepithelioma-like carcinoma of the skin (LELCS)**
4665. **Primary mucinous carcinoma of the skin**
4666. **Primary Thrombotic Microangiopathy**
4667. **Proctalgia fugax**
4668. **Autoimmune progesterone dermatitis AIPD**
4669. **Progesterone-induced stomatitis**
4670. **Werner's syndrome=progeria of the adult=pangeria**
4671. **Progeroid syndrome, petty type**
4672. **Hutchinson–Gilford progeria syndrome =progeria Infantum**
4673. **progressive cribriform and zosteriform hyperpigmentation PCZH**
4674. **Progressive macular hypomelanosis PMH**
4675. **Progressive osseus heteroplasia**
4676. **Progressive papular purpuric eruption**
4677. **Progressive Zosteriform Macular Pigmented Lesion (PZMPL)**
4678. **Prolidase deficiency(PD)=hyperimidodipeptiduria**
4679. **Proliferating pilar tumor**
4680. **Proliferative fasciitis**
4681. **prominent platysmal bands**
4682. **Propionic acidemia**
4683. **Proteasome- associated autoinflammatory syndrome (PRAAS)**
4684. **Protein contact dermatitis**
4685. **Protein C deficiency**
4686. **Protein S deficiency**
4687. **Proteus syndrome PS = Cerebriform fibrous proliferation**
4688. **Elattoproteus syndrome**

4689. **Protothecosis**
4690. **Prune belly syndrome**
4691. **Prurigiform angiomatosis (PA)**
4692. **Prurigo = Acne urticata**
4693. **Prurigo besnier**
4694. **Prurigo hebra**
4695. **Prurigo gravidarum**
4696. **Prurigo nodulris**
4697. **Prurigo pigmentosa**
4698. **Prurigo of pregnancy**
4699. **Prurigo simplex**
4700. **Actinic prurigo = Hutchinson's summer prurigo =Hydroa Aestivale**
4701. **Pruritic and dyskeratotic dermatosis**
4702. **Pruritus in scars/Keliod**
4703. **Aquagenic Pruritus**
4704. **Contagious Pruritus**
4705. **Pruritus ani**
4706. **Pruritus scroti**
4707. **Pruritus vulvae**
4708. **Renal Pruritus**
4709. **Scalp Pruritus**
4710. **Hematologic Pruritus**
4711. **Postmenopausal Pruritus**
4712. **Pruritus in Internal Malignancy**
4713. **Cholestatic Pruritus**
4714. **Senile Pruritus**
4715. **Endocrine pruritus**
4716. **Referred Pruritus (Mitempfindung)**
4717. **Psychogenic pruritus= Somatoform Pruritus**
4718. **Pharmacologic pruritus**
4719. **Pseudallescheriasis**

Pseudo in dermatology (see next)

4720. ✓ Pseudo hypoparathyroidism
4721. ✓Pseudo pseudo hypoparathyroidism
4722. ✓ Pseudoichthyosis, acquired
4723. ✓ Eruptive pseudoangiomatosis
4724. ✓ Pseudo acanthosis nigricans
4725. ✓ Pseudo acne of nasal crease
4726. ✓ Pseudo ainhum
4727. ✓ Pseudo albinism
4728. ✓ Pseudo alopecia areata
4729. ✓ Pseudo behcet's disease
4730. ✓ Pseudo blue-lunula
4731. ✓ Pseudo botryomycosis
4732. ✓ Pseudo bowen's disease
4733. ✓ Pseudo bubo
4734. ✓ Pseudo chancre redux
4735. ✓ Pseudo chromhidrosis
4736. ✓ Pseudo darier's sign
4737. ✓ Pseudo dermatomyositis
4738. ✓ Pseudo furuncle
4739. ✓Pseudoglucagonoma
4740. ✓ Pseudo gout
4741. ✓ Pseudo halo nevus
4742. ✓ Pseudo horn cyst
4743. ✓ Pseudo hutchison's sign
4744. ✓ Pseudo lupus vulgaris
4745. ✓ Pseudo melanoma
4746. ✓ Pseudo monilethrix
4747. ✓ Pseudo mycetoma
4748. ✓ Pseudo mycosis fungoides
4749. ✓ Pseudo ochronosis

4750. ✓ Pseudo rheumatoid nodule
4751. ✓ Pseudosarcoma
4752. ✓ Pseudo scabies
4753. ✓ Pseudo scleroderma
4754. ✓ Pseudo sign of leser trelat
4755. ✓ Pseudo sporotrichosis
4756. ✓ Pseudoacromegaly
4757. ✓ Pseudocellulitis
4758. ✓ Pseudocyst of the auricle
4759. ✓ Pseudodidymosis aplasticosebacea
4760. ✓ Pseudoepitheliomatous hyperplasia
4761. ✓ Pseudofolliculitis barbae =pili recurvate= pili incarnati
4762. ✓ Pseudofolliculitis corporis
4763. ✓ Pseudofolliculitis vibrissae
4764. ✓ Pseudomamma
4765. ✓ Pseudomyogenic hemangioendothelioma
4766. ✓ Pseudomelanocytic nests
4767. ✓ Pseudopelade of brocq
4768. ✓ Pseudopili annulati
4769. ✓ Pseudotumor cerebri
4770. ✓ Pseudo kaposi
4771. ✓ Pseudo knuckle pad (chewing pad)
4772. ✓ Pseudo-koenen tumors
4773. ✓ Pseudomacrolunula
4774. ✓ Pseudo-pseudoxanthoma elasticum
4775. ✓ Pseudo-pseudo hutchison's sign
4776. ✓ Pseudovasculitis
4777. Pseudoepitheliomatous keratotic and micaceous balanitis
4778. Pseudoverrucous papule and nodule
4779. Perianal pseudoverrucous papules and nodules (PPPN)

4780. **Pseudoxanthoma elasticum PXE= Grönblad-strandberg syndrome= systematized elastorrhexis**
4781. **Pseudoxanthoma elasticum-like papillary dermal elastolysis**
4782. **Pseudomonaus related diseases (see next)**
4783. ✓ **Pseudomonas hand foot syndrome**
4784. ✓ **Ecthyma gangrenosum**
4785. ✓ **Green nail syndrome**
4786. ✓ **Pseudomonal pyoderma**
4787. ✓ **Blastomycosis-like pyoderma**
4788. ✓ **Otitis externa**
4789. ✓ **pseudomonal folliculitis -hot tub folliculitis**
4790. ✓ **Malignant otitis externa**
4791. ✓ **Pseudomona chondritis**
4792. ✓ **Septicemia**
4793. **Psoriasis vulgaris=Chronic Plaque Psoriasis**
4794. **Annular Psoriasis**
4795. **Elephantine Psoriasis**
4796. **Erythrodermic Psoriasis**
4797. **Follicular Psoriasis**
4798. **Guttate Psoriasis**
4799. **Inverse Psoriasis**
4800. **Lichenoid psoriasis**
4801. **Linear Psoriasis**
4802. **Ostraceous Psoriasis**
4803. **Psoriasis :Acrodermatitis continua of hallopeau**
4804. **Psoriasiform acral dermatitis**
4805. **Psoriasiform keratosis**
4806. **Psoriasiform napkin dermatitis**
4807. **Psoriasis alba**
4808. **Psoriasis herpeticum**
4809. **Psoriasis punctate= micro-guttate**
4810. **Psoriasis vaccinalis**

4811. **Psoriatic arthritis**
4812. **Psoriatic neurodermiformis**
4813. **Psoriatic onycho-pachydermo-periostitis (POPP)**
4814. **Pustular Psoriasis**
4815. **Generalized pustular psoriasis of pregnancy**
4816. **Pustulosis palmaris et plantaris**
4817. **Rupioid Psoriasis**
4818. **Sebopsoriasis**
4819. **Verrucous psoriasis**
4820. **Von zumbusch pattern of generalized Pustular Psoriasis**
4821. **Psema (entity with clinical features of both psoriasis and eczema)**
4822. **PSTPIP1-associated myeloid-related-proteinemia inflammatory syndrome (PAMI Syndrome)**
4823. **Psychogenic pruritus**
4824. **Psychogenic purpura =Gardner-diamond syndrome=Autoerythrocyte sensitization = painful bruising syndrome**
4825. **Pten hamartoma tumor syndrome (PHTS)**
4826. **Linear PTEN nevus (linear cowden nevus included)**
4827. **Pterygium unguis**
4828. **Puffy hand syndrome**
4829. **Puffy foot syndrome**
4830. **Pulicosis= Puliciasis**
4831. **Puretic syndrome**
4832. **Purpura**
4833. **Contact purpura**
4834. **Purpura cyclops**
4835. **Purpura en cocarde**
4836. **Purpura factitia**
4837. **Purpura fulminans**
4838. **Purpura gogglorum**
4839. **Postictal Thoracocervicofacial Purpura**
4840. **Purpura progressiva pigmentosa**
4841. **Purpura pulicosa**

4842. **Purpura simplex**
4843. **Solar purpura=Solar capillaritis**
4844. **Purpura telangiectatica arciformis**
4845. **Pinch purpura**
4846. **Pustular vasculitis**
4847. **Pustular Vasculitis of the hands PVH**
4848. **Pustulosis acuta generalisata (PAG)**
4849. **Pustulotic arthro-osteitis (PAO)=Sonozaki syndrome**
4850. **Puva (psoralen and UVA) related diseases**
4851. **Puva-induced acrobullous dermatosis**
4852. **Puva induced keratosis**
4853. **Puva induced lentigines**
4854. **pyemotes ventricosus dermatitis**
4855. **Pyknodysostosis**
4856. **Pyoderma faciale**
4857. **pyoderma fistulan=fox den disease**
4858. **Pyoderma, folliculitis and atopy, with defective leukocyte and lymphocyte function**
4859. **Pyoderma gangrenosum (see next)**
4860. ✓ **Ulcerative**
4861. ✓ **Vesiculobullous**
4862. ✓ **Pustular**
4863. ✓ **Superficial granulomatous**
4864. ✓ **Pyostomatitis vegetans**
4865. ✓ **Pyodermatitis-pyostomatitis vegetans**
4866. **Pyogenic granuloma (lobular capillary haemangioma = lobular capillary hemangioma =Botryomycoma= granuloma telangiectaticum= Granuloma pyogenicum**
4867. **Warner and wilson-jones syndrome (recurrent pyogenic granuloma with satellite lesions)**
4868. **Agminated pyogenic granulomas**
4869. **Congenital Disseminated Pyogenic Granuloma**

4870. **Eruptive pyogenic granuloma**
4871. **Pyogenic granuloma-like kaposi sarcoma (PG-like ks)**
4872. **Pyogenic granuloma ike metastases**
4873. **Pregnancy-Related Pyogenic Granuloma (Granuloma Gravidarum)**
4874. **Pyogenic infections**
4875. **Pyomyositis**
4876. **Pyramidal protrusion=Perineal skin tag**
4877. **Pyrin-associated autoinflammation with neutrophilic dermatosis**
4878. **Pyruvoyltetrahydropterin synthase (6-ptps) deficiency**
4879. **Pythiosis**
4880. **Q fever**
4881. **Quincke sign**
4882. **Rabson -Mendenhall syndrome**
4883. **Rac2 deficiency**
4884. **RACAND (Raynaud's phenomenon, anticentromere antibodies,digital necrosis,without digital sclerosis)**
4885. **Racoon eyes**
4886. **Radiation dermatitis (Acute&Chronic)**
4887. **Radiotherapy-induced oral mucositis**
4888. **Radiation recall dermatitis**
4889. **Radiation-induced comedones**
4890. **Radiation-Induced Atypical Vascular Lesion**
4891. **Fluoroscopy-induced radiation dermatitis**
4892. **Ranula**
4893. **Rat-bite fever=erythema arthriticum epidemicum=Sodoku=Haverhill fever**
4894. **Ramsay–hunt syndrome**
4895. **RASopathies**
4896. **Rautenstrauch syndrome**
4897. **Raynaud phenomenon**
4898. **Raynaud syndrome**
4899. **Reactive angioendotheliomatosis**
4900. **Malignant angioendotheliomatosis**
4901. **Reactive arthritis**

4902. **Reactive infectious mucocutaneous eruption**
4903. **Reactive perforating collagenosis. RPC**
4904. **Recalcitrant, erythematous, desquamating (RED) disorder**
4905. **Recurrent cutaneous eosinophilic vasculitis**
4906. **Recurrent Neutrophilic Dermatosis of the Face**
4907. **Recurrent respiratory papillomatosis**
4908. **Recurrent toxin-mediated perineal erythema**
4909. **Recurring digital fibrous tumors of childhood (benign juvenile digital fibromatosis)**
4910. **Red baby syndrome**
4911. **Red ear syndrome**
4912. **Red finger syndrome**
4913. **Red man syndrome**
4914. **Red neck syndrome**
4915. **Red scrotum syndrome**
4916. **Reed syndrome (Hereditary Leiomyomatosis and Renal Cell Cancer Syndrome)**
4917. **Reiter syndrome=Circinate balanitis=keratoderma blennorrhagicum= reactive arthritis=circinate ulcerative vulvitis**
4918. **Relapsing eosinophilic perimyositis**
4919. **Relapsing linear acantholytic dermatosis**
4920. **Relapsing polychondritis (RPC)**
4921. **Remitting seronegative symmetrical synovitis with pitting edema (RS3PE) syndrome**
4922. **Restrictive dermopathy=tight skin contracture syndrome**
4923. **Reticular dysgenesis**
4924. **Reticular/reticulates telangiectatic erythema**
4925. **Reticular erythema with focal mid-dermal elastophagocytosis (REMDE)**
4926. **Reticulate acropigmentation of kitamura**
4927. **Reticulate hyperpigmented scleroderma RHS**
4928. **Reticulated acanthoma with sebaceous differentiation**
4929. **Reticulated acropigmentation of Dohi**
4930. **Reticulate Genital Melanosis with Localized Depigmentation**

4931. **Reticulate vascular nevus =Cutis marmorata telangiectatica congenita, congenital livedo reticularis**
4932. **Retiform hemangioendothelioma**
4933. **Restless legs syndrome**
4934. **Revesz syndrome**
4935. **Reye syndrome**
4936. **Malignant rhabdoid tumor MRT**
4937. **Rhabdomyoma**
4938. **Rhabdomyomatous mesenchymal hamartoma=striated muscle Hamartoma**
4939. **Rhabdomyosarcoma**
4940. **Botryoid embryonal rhabdomyosarcoma**
4941. **Rheumatic fever nodule**
4942. **Rheumatoid arthritis &Rheumatoid nodule & Bywatery lesions & Felty syndrome**
4943. **Rheumatic neutrophilic dermatosis RND**
4944. **Rhinophyma**
4945. **Rhinoscleroma**
4946. **Rhinosporidiosis**
4947. **Rhizopus fungal infection**
4948. **Rhytids**
4949. **Riboflavin deficiency= Vitamin B2 deficiency**
4950. **Richter syndrome**
4951. **Rickettsia aeschlimannii infection**
4952. **Rickettsia parkeri rickettsiosis**
4953. **Rickettsia massiliae rickettsiosis**
4954. **Rickettsial pox**
4955. **Rickettsia species 364D rickettsiosis**
4956. **Riehl's melanosis = pigmented contact dermatitis**
4957. **Riga fede disease**
4958. **Robert syndrome with tetraphacomelia= pseudothalidomide syndrome**
4959. **Rocky mountain spotted fever**
4960. **Rogue hairs**
4961. **Rombo syndrome**

4962. **Rosacea (Erythematotelangiectatic & Papulopustular & Phymatous & Ocular & Granulomatous)**
4963. **Rosacea fulminans=pyoderma faciale**
4964. **Rosacea like lupus erythematosus**
4965. **Rosacea like tinea facei**
4966. **Rosacea-like demodicidosis of the scalp**
4967. **Rosacea conglobate**
4968. **Halogen rosacea**
4969. **Gram negative rosacea**
4970. **Rosenthal-kloepfer syndrome**
4971. **Rosselli–gulienetti syndrome**
4972. **Roseola infantum= exanthem subitum= 6th disease sixth= Pseudorubella**
4973. **Roseola vaccinatum**
4974. **Ross River virus infection**
4975. **Ross syndrome**
4976. **Rose handler's disease**
4977. **Rothmann-Makai syndrome**
4978. **Rothmund-Thomson syndrome =poikiloderma congenitale**
4979. **Roth-bernhardt syndrome**
4980. **Rowell syndrome**
4981. **Rubella virus-associated granulomatous disease (RuV-GD)**
4982. **Rubella= German measles=Three-day measles=Third Disease**
4983. **Rubinstein–taybi syndrome=Broad thumb–hallux syndrome**
4984. **Rud syndrome**
4985. **Rüdiger syndrome**
4986. **Rudimentary toe**
4987. **Ruggieri-Happle syndrome**
4988. **Rumpel Leede sign**
4989. **Ruocco's immunocompromised cutaneous district**
4990. **Russell–Silver syndrome**
4991. **Russel sign**
4992. **Sabinas brittle hair and mental deficiency syndrome**

4993. **Salivary gland neoplasms (Pleomorphic adenoma&Canalicular adenoma &Mucoepidermoid carcinoma&Polymorphous adenocarcinoma&Adenoid cystic carcinoma)**
4994. **Salt and pepper leukoderma**
4995. **Saddan syndrome**
4996. **Saddle nose**
4997. **Saethre-chotzen syndrome**
4998. **Sago palm disease**
4999. **SAHA syndrome :seborrhea, acne, hirsutism, and alopecia**
5000. **Sakati syndrome**
5001. **Salamon syndrome**
5002. **SAM syndrome (severe dermatitis–multiple allergies–metabolic wasting)**
5003. **SAMD9L-associated autoinflammatory disorder (SAAD)**
5004. **Sandmann–Andra syndrome**
5005. **Sandfly fever =phlebotomus fever**
5006. **Sarcoidosis (see next)**
5007. **✓Papules**
5008. **✓ plaques**
5009. **✓ lupus pernio**
5010. **✓ Scar**
5011. **✓ Tattoo**
5012. **✓Lichenoid**
5013. **✓Alopecic**
5014. **✓ Annular**
5015. **✓ Atrophic**
5016. **✓ lichenoid**
5017. **✓Psoriasiform**
5018. **✓ Subcutaneous (Darier-Roussy)**
5019. **✓ Angiolupoid**
5020. **✓ Erythrodermic**
5021. **✓ Hypopigmented**
5022. **✓ Ichthyosiform**

5023. ✓ Micropapular
5024. ✓ Ulcerative
5025. ✓ Verrucous
5026. ✓ Morpheaform
5027. ✓ Sarcoidois early onset type
5028. ✓ Pseudotumoral cutaneous sarcoidosis
5029. Sarcoma
5030. Sarcomata
5031. Scabies **(see next)**
5032. ✓ Crusted / Norwegian scabies
5033. ✓ Bullous scabies
5034. ✓ Infantile scabies
5035. ✓ Scabies inclean
5036. ✓ Scabetic nodule
5037. ✓ Pseudo scabies
5038. ✓ Pyoscabies=infected scabies
5039. ✓ Scabies surrepticius
5040. ✓ Scabies herpeticum
5041. ✓ Scabies incognito
5042. ✓ Scabid
5043. Saurian papulosis
5044. Scalp–ear–nipple syndrome (Finlay–Marks syndrome)
5045. Scarlet fever=Scarlatina
5046. SCALP syndrome (sebaceous nevus-cns malformations-aplasia cutis congenital-limbal dermoid-pigmented nevus)
5047. Scalp dysesthesia
5048. Hypertrophic scar
5049. Atrophic scar
5050. Simple scar
5051. Scars of prematurity
5052. SCARF syndrome(skeletal abnormalities, cutis laxa, craniostenosis ,ambiguous genitalia, retardation, facial abnormalities)

5053. **Scedosporiosis**
5054. **Schimke immuno-osseous dysplasia (**
5055. **Schimmelpenning–feuerstein–mims syndrome SFM**
5056. **Schindler disease (α -Nacetylgalactosaminidase =α - NAGA deficiency)**
5057. **Schinzel–Giedion midface retraction syndrome**
5058. **Schnitzler's syndrome**
5059. **Schöpf–Schulz–Passarge syndrome**
5060. **Schwannoma = Neurilemmoma**
5061. **Neuroblastoma-Like Schwannoma**
5062. **Plexiform Schwannoma**
5063. **Hybrid schwannoma /perineurioma**
5064. **Nodular scleroderma**
5065. **Scleredema**
5066. **Scleredema of Bushke**
5067. **Sclerema neonatorum**
5068. **Scleroderma**
5069. **Scleroderma-like cutaneous metastasis**
5070. **Sclerodermatomyositis**
5071. **Sclerodermoid disorders**
5072. **Scleromalacia**
5073. **Scleromyositis**
5074. **Scleromyxedema**
5075. **Sclerosing epithelioid fibrosarcoma**
5076. **Sclerosing lipogranuloma**
5077. **Systemic Sclerosis**
5078. **Diffuse Systemic sclerosis**
5079. **Limited Systemic sclerosis**
5080. **Systemic Sclerosis Overlap Syndromes**
5081. **Intermediate cutaneous scleroderma (icSSc)**
5082. **Systemic Sclerosis Sine scleroderma SSc (ssSSc)**
5083. **Sclerotic fibroma=Storiform Collagenoma**
5084. **Scrotal cyst /idiopathic scrotal calcification**
5085. **Scrotal fat necrosis**

5086. **Scrub typhus= Tsutsugamushi disease**
5087. **Scurvy**
5088. **Scytalidium infections**
5089. **Seaweed dermatitis**
5090. **Sea urchin granuloma**
5091. **Sealpox**
5092. **Sebastian syndrome**
5093. **Sebaceous casts**
5094. **Sebaceous filaments**
5095. **Hyperactive Sebaceous glands of newborn**
5096. **Sebaceoma**
5097. **Sebaceous carcinoma**
5098. **Sebaceous epithelioma**
5099. **Sebaceous hyperplasia**
5100. **Sebaceous gland prominence**
5101. **Sebaceous hyperplasia variant: juxtaclavicular beaded lines= Striated beaded lines**
5102. **Sebaceous adenoma**
5103. **Seboacanthoma**
5104. **Sebocystomatosis**
5105. **Sebofolliculoma**
5106. **Sebomatricoma**
5107. **Seborrheic dermatitis =Seborrhoeic eczema= Seborrhea**
5108. **Petaloid seborrheic dermatitis**
5109. **Seborrhoea Herpeticum**
5110. **Seborrheic keratosis SK (Pedunclated & Pigmented & Superficial & Eruptive & Irritated&Tadpoleseborrheic keratosis)**
5111. **SASKIA nevus (segmentally arranged seborrheic keratoses with impending atypia**
5112. **Seabather's Eruption=Sea lice**
5113. **Seborrheic melanosis**
5114. **Seckel syndrome**
5115. **Secretan syndrome**

5116. **Secretory carcinoma of the skin**
5117. **Segmental congenital vascular anomaly with atrophy, ulceration and scarring (SeCVAUS)**
5118. **Segmental Pigmentation Disorder**
5119. **Selective IGA deficiency**
5120. **Selective IGM deficiency**
5121. **Selenium toxicity/excess**
5122. **Selenium Deficiency= Keshan disease=Kashin–Beck disease**
5123. **Sener syndrome (frontonasal dysplasia and dilated virchow –Robin spaces)**
5124. **Senile gluteal dermatosis**
5125. **Senile purpura**
5126. **Sennetsu fever**
5127. **Sensitive skin syndrome**
5128. **Sensorineural hearing loss, enamel hypoplasia and nail defects (Heimler syndrome)**
5129. **Septic Vasculopathy**
5130. **Seroma= lymphocoele**
5131. **Serpentine supravenous hyperpigmentation**
5132. **Serratia marcescens**
5133. **Servelle Martorell syndrome=Port-wine stain, venous malformations and limb hypotrophy**
5134. **Setleis' syndrome**
5135. **Severe Achondroplasia With Developmental Delay and Acanthosis Nigricans (SADDAN)**
5136. **Severe combined immunodeficiency SCID (X-linked SCID &JAK3 deficiency & Interleukin-7 receptor deficiency &ADA deficiency &PNP deficiency &Reticular dysgenesis &Coronin 1A deficiency &Omenn syndrome &Artemis deficiency&PRKDC deficiency&LIG4 syndrome&NHEJ1 syndrome&TCR deficiencies &CD45 deficiency&ZAP-70 deficiency & CD8 deficiency&Calcium channel deficiency&MHC class I deficiency &MHC class II deficiency&Winged helix nude deficiency)**
5137. **Severe dermatitis, multiple allergies, and metabolic wasting (SAM)**

5138. **Sexual abuse**
5139. **Sexual Transmitted diseases STDs**
5140. **Shaheen syndrome**
5141. **Shitake dermatitis**
5142. **Short anagen syndrome**
5143. **Short stature with pleonosteosis and periarticular fibrosis = léri's syndrome**
5144. **Shprintzen–Goldberg syndrome**
5145. **Shwachman-Diamond syndrome**
5146. **Sialidosis**
5147. **Sialolith**
5148. **Sideroblastic anemia with B cell immunodeficiency, periodic fevers and developmental delay**
5149. **Siderosis**
5150. **Silica granuloma**
5151. **Silicone granuloma= Siliconoma**
5152. **Silver nirate pigmentation**
5153. **Simpson-Golabi-Behmel syndrome**
5154. **Sindbis virus infection**
5155. **Singleton-Merten syndrome (SMS)**

"sines" in dermatology (see next)

5156. ✓ **Acne fulminans sine fulminans**
5157. ✓ **Dermatomyositis sine myositis**
5158. ✓ **Eccrine hidradenitis sine neutrophils**
5159. ✓ **Eruptive seborrheic keratosis sine malignancy**
5160. ✓ **linear melorheostotic scleroderma with hypertrichosissine melorheostosis**
5161. ✓ **Lupus sine lupo**
5162. ✓ **Malignant acanthosis nigricans sine malignancy**
5163. ✓ **Necrobiosis lipoidica sine diabetes**
5164. ✓ **Pellagra sine pellagra**
5165. ✓ **Pruritus sine materia**
5166. ✓ **Psoriatic arthritis sine psoriasis**

5167. ✓ **Systemic sclerosis sine scleroderma**
5168. ✓ **Zoster sine herpete**
5169. **Sinusoidal hemangioma**
5170. **Sinus pericranii**
5171. **Sipple's syndrome**
5172. **Sirenomelia (Mermaid syndrome)**
5173. **Sister Marry joseph nodule**
5174. **Sitosterolemia**
5175. **Sjogren syndrome**
5176. **Skeeter syndrome**
5177. **Skin-ache syndrome**
5178. **Skin fragility syndrome**
5179. **SKINTED (Surgery of the Knee, Injury to the Infrapatellar branch of the saphenous Nerve, Traumatic Eczematous Dermatitis)**
5180. **Slapped cheek syndrome**
5181. **Slapped neck solar melanosis**
5182. **Smith–Lemli–Opitz syndrome**
5183. **Small pox infection=Variola major**
5184. **Smegma pearls**
5185. **Smooth muscle hamartoma SMH=Congenital arrector pili Hamartoma= Muscle naevi**
5186. **Sneddon Wilkinson disease**
5187. **Sneddon syndrome= Idiopathic livedo reticularis with systemic involvement**
5188. **Sock line bands**
5189. **Sock line hyperpigmentation**
5190. **SOFT syndrome (short stature, onychodysplasia, facial dysmorphism, and hypotrichosis)**
5191. **Solar /Actinic elastosis**
5192. **Solar elastotic bands**
5193. **Solamen syndrome**
5194. **Solitary fibrous tumor**
5195. **Sotos syndrome**

5196. **Sparganosis**
5197. **Specific granule deficiency**
5198. **Spider angioma= Spider nevus= Nevus araneus=Spider telangiectasia**
5199. **Spiegler–Fendt sarcoid -lymphocytoma cutis**
5200. **Spinal dysraphism**
5201. **Spindle cell hemangioma**
5202. **Spindle hemangioendothelioma**
5203. **Spindle cell lipoma**
5204. **Spiradenoma**
5205. **Spiradenocylindroma**
5206. **Spiradenocylindrocarcinoma**
5207. **Spiradenocarcinoma**
5208. **Malignant eccrine spiradenoma (MES)**
5209. **Split hand -foot malformation (SHFM1)**
5210. **Spondyloenchodrodysplasia with immune dysregulation**
5211. **Spontaneous scrotal ulceration**
5212. **Sporobolomyces salmonicolor**

Spots in dermatology (see next)

5213. ✓ **Nickel spot test**
5214. ✓ **Cotton wool spots**
5215. ✓ **Stasis spots**
5216. ✓ **Antimony spots**
5217. ✓ **Ash leaf spots**
5218. ✓ **Bier spots**
5219. ✓ **Bitot's spots**
5220. ✓ **Black spot test**
5221. ✓ **Black‐spot poison ivy**
5222. ✓ **Brushfield spots**
5223. ✓ **Café au lait spots**
5224. ✓ **Café noir spot**
5225. ✓ **Campbell de morgan spots**
5226. ✓ **Cayenne pepper spots**

5227. ✓Drusen‐like spots
5228. ✓Forchheimer spots
5229. ✓Fordyce spots
5230. ✓Herman spots
5231. ✓Ink spot lentigo=sunburn lentigo
5232. ✓Janeway spots
5233. ✓Koplik spots
5234. ✓Leopard skin spotting
5235. ✓Liver spots
5236. ✓Lucio's spotted leprosy
5237. ✓Milk spots
5238. ✓Mongolian spot
5239. ✓Monospot test
5240. ✓Nagayama spots
5241. ✓Oil spots
5242. ✓Orange palpebral spots
5243. ✓Rose spots
5244. ✓Roseolar spots
5245. ✓Roth spots
5246. ✓Shin spots (binkley's spots, pigmented pretibial patches, spotted leg syndrome, diabetic dermangiopathy)
5247. ✓Sporty spots
5248. ✓Spot urine tests
5249. ✓Spot tests in allergic contact dermatitis
5250. ✓Spotted fevers
5251. ✓Spotted lunula
5252. ✓Spotty parakeratosis=staccato parakeratosis:
5253. ✓Twin spots (syn: didymosis)
5254. ✓White spot disease (syn: lichen sclerosus)
5255. Sporotrichosis
5256. Squamous cell papilloma

5257. **Squamo-melanocytic tumor**
5258. **Squamous metaplasia of the lactiferous ducts**
5259. **Squamous cell carcinoma**
5260. **Verrocous squamous cell carcinoma**
5261. **Squamoid eccrine ductal carcinoma**
5262. **Staphylococal scalded skin syndrome SSSS=Ritter disease=pemphigus neonatorum**
5263. **Stasis purpura**
5264. **Stasis dermatitis**
5265. **Steal syndrome**
5266. **Steatocystoma (simplex & multiplex & multiplex suppurativum)**
5267. **Sternal cleft**
5268. **Stellate scar**
5269. **Steriod induced rosacea**
5270. **Stevens-johnson syndrome SJS**
5271. **SJS / TEN overlap**
5272. **Fuchs' syndrome (stevens-johnson syndrome without skin involvement)**
5273. **Stewart-Bluefarb syndrome**
5274. **Stewart-Treves syndrome=Postmastectomy lymphangiosarcoma**
5275. **Stickler syndrome**
5276. **Stiff baby syndrome**
5277. **Stiff skin syndrome =Congenital fascial dystrophy**
5278. **Adult-onset Still disease**
5279. **Systemic-onset juvenile idiopathic arthritis (sJIA)= Still disease**
5280. **Sting-associated vasculopathy with onset in infancy SAVI**
5281. **STK4 deficiency**
5282. **Stomal related diseases**
5283. **Stomatitis medicamentosa**
5284. **Gold-induced stomatitis**
5285. **Allergic Contact Stomatitis**
5286. **Antibiotic-induced stomatitis**
5287. **Storiform collagenoma**
5288. **Storm's syndrome**

5289. **Straight hair nevus**
5290. **Strawberry in dermatology (see next)**
5291. ✓ **Strawberry gingival hyperplasia**
5292. ✓ **Strawberry cervix**
5293. ✓ **Strawberry nevus**
5294. ✓ **Strawberry papillomatosis**
5295. ✓ **Strawberry like nasal growth**
5296. ✓ **Strawberry tongue**
5297. **Streptocerciasis**
5298. **Streptococcal intertrigo**
5299. **Streptococcal sex syndrome**
5300. **Striae = strech mark= Atrophoderma maculatum**
5301. ✓ **Striae Alba**
5302. ✓ **Striae Rubra**
5303. ✓ **Striae Distensae**
5304. ✓ **Striae Gravidarum**
5305. **Striated muscle hamartoma**
5306. **Strongyloidiasis**
5307. **Stucco keratosis**
5308. **Sturge weber syndrome (Encephalotrigeminal angiomatosis)**
5309. **Subaponeurotic (subgaleal) fluid collection**
5310. **Subcorneal pustular dermatosis= Sneddon–Wilkinson disease**
5311. **Subcutaneous emphysema**
5312. **Subcutaneous fat necrosis of the newborn**
5313. **Subepidermal calcified nodule**
5314. **Subgaleal empyema**
5315. **Subgaleal hematomas**
5316. **Sucking blisters**
5317. **Suction Blisters**
5318. **Suction purpura/factitious purpura**
5319. **Sudden whitening of hair**
5320. **Sudoriparous angiomas SA**

5321. **Sulzberger–Garbe syndrome= Exudative discoid and lichenoid chronic dermatosis= Oid-oid disease**
5322. **Sunburn= Dermatitis solaris**
5323. **Superficial acral fibroblastic spindle cell neoplasm with EWSR1-SMAD3 fusion**
5324. **Superficial CD34-positive fibroblastic tumor**
5325. **Superficial migratory thrombophlebitis**
5326. **Superficial papular neuroma**
5327. **Superior vena cava syndrome**
5328. **Supernumeray nostril**
5329. **Supravenous lichen planus**
5330. **Sweat: Hyperhidrosis &Hypohidrosis &Anhidrosis**
5331. **Sweet syndrome : Acute Febrile neutrophilic dermatosis**
5332. **Swimmer's ear**
5333. **Swimmer's itch =Cercarial dermatitis= Dermatitis schistosomica**
5334. **Swimmers's xerosis**
5335. **Sycosis Barbae=Sycosis Vulgaris= Barber's Itch**
5336. **Mycotic Sycosis**
5337. **Herpetic Sycosis**
5338. **Lupoid Sycosis**
5339. **Symmetric acral annular erythema**
5340. **Symmetric idiopathic micropapular facial eruption (SIMPLE)**
5341. **Symmetrical acrokeratoderma (SAK)**
5342. **Symmetrical palatal fibromatosis**
5343. **Symmetrical peripheral gangrene**
5344. **Symmetrical progressive leukopathy**
5345. **Symmetric synovitis**
5346. **Symplastic hemangioma**
5347. **Synovial cyst**
5348. **Synovial metaplasia**
5349. **Synovial sarcoma**
5350. **Synovitis, acne, pustulosis, hyperostosis and osteitis SAPHO syndrome**

5351. **Syphilis = great pox (primary chancre& secondary & &congenital& lues maligna)**
5352. **Secondary syphilis : (collarette of biett & condylomata lata & corona veneris & necklace of venus & syphilitic perlèche & plaques muqueuses)**
5353. **Tertiary syphilis (late): gummas**
5354. **Syphilitic balanitis of follmann (SBF)**
5355. **Malignant syphilis=Lues maligna of syphilis**
5356. **Congenital syphilis (rhagades & hutchinson teeth & mulberry molars & saddle nose &olympian brow& higouménakis' sign &saber shins & clutton's joints)**
5357. **Herpetic syringitis**
5358. **Syringoacanthoma of rahbari**
5359. **Syringocystadenoma papilliferum SCAP**
5360. **Syringocystadenocarcinoma papilliferum SCAP**
5361. **Syringoma (Clear&Localized &Eruptive& Familial)**
5362. **Eruptive syringomas =Eruptive hidradenoma of Darier and Jaquet**
5363. **Eruptive milium-like syringoma (EMS)**
5364. **Infiltrating Syringomatous Adenoma of Nipple**
5365. **Syringofibroadenoma= Acrosyringeal nevus of weedon and lewis**
5366. **Syringoid eccrine carcinoma= Eccrine carcinoma**
5367. **Syringomatous carcinoma**
5368. **Syringolymphoid Hyperplasia with alopecia and anhidrosis**
5369. **Syringomyelia**
5370. **Skin tag = Acrochordon = Fibroepithelial polyp =Soft fibroma=Lax Fibroids**
5371. **Takayasu arteritis**
5372. **Tangier disease =Alpha-Lipoprotein Deficiency Disease**
5373. **Tail /faun tail**
5374. **Talaromyces marneffei infection**
5375. **Talon noir**
5376. **Tanapox**
5377. **Tapered fracture hair**
5378. **TAR syndrome (Thrombocytopenia with absent radius syndrome)**
5379. **Tattoo**

5380. **Taurodontia, absent teeth and sparse hair**
5381. **Tay syndrome**
5382. **Teebi–Shaltout syndrome**
5383. **Telangiectasia**
5384. **Eruptive recurrent pigmented telangiectasia**
5385. **Telangiectasia macularis eruptive perstan TMEP**
5386. **Telangiectasia macularis multiplex acquisita**
5387. **Telangiectatic pacemaker erythema TPE**
5388. **Telogen effluvium (Acute telogen effluvium&Chronic diffuse hair loss&Primary chronic telogen effluvium)**
5389. **TEMPI syndrome (Telangiectasias, Erythrocytosis , Monoclonal gammopathy, Perinephric-fluid collections , and Intrapulmonary shunting)**
5390. **Tennis toe=Runner's toe=Jogger's toe**
5391. **Teratoma**
5392. **Terminal osseous dysplasia with pigmentary defects TODPD**
5393. **Terra firma- forme dermatosis TFFD**
5394. **Tetramelic deficiencies, ectodermal dysplasia, deformed ears and other abnormalities (odontotrichomelic syndrome)**
5395. **Texier disease**
5396. **Thanos syndrome**
5397. **Thinker sign**
5398. **Thromboangitis obliterans**
5399. **Thrombocytopenic purpura**
5400. **Thrombotic thrombocytopenic purpura**
5401. **ICI-(Immune Checkpoint Inhibitor)induced thrombotic vasculopathy**
5402. **Thumb- and finger sucking**
5403. **Thumb deformity-alopecia-pigmentation anomaly syndrome**
5404. **Thymic dysplasia**
5405. **Thymic cyst**
5406. **Thyroglossal cyst**
5407. **Tibialis anterior muscle herniation**
5408. **Tick borne lymphadenopathy (TIBOLA)**
5409. **Tick-borne relapsing fever**

5410. **Tietz syndrome**
5411. **Tinea amiantacea =pityriasis amiantacea**
5412. **Tinea barbae**
5413. **Tinea blepharociliaris**
5414. **Tinea capitis (scaly & black dot& grey patch&pustular& kerion& agminate folliculitis)**
5415. **Tinea favosa= favus**
5416. **Tinea corporis= Ringworm of the body=Tinea circinata**
5417. **Tinea corporis bullosa**
5418. **Tinea corporis hyperkeratotica**
5419. **Tinea cruris= Ringworm of the groin**
5420. **Tinea gladiatorum**
5421. **Tinea faciei = Ringworm of the face**
5422. **Tinea incognito**
5423. **Tinea imbricata**
5424. **Tinea pseudoimbricata**
5425. **Majocchi granuloma MG =Granuloma tricofitico =Nodular granulomatous perifolliculitis**
5426. **Tinea manuum**
5427. **Tinea nigra= Keratomycosis mgncans palmaris**
5428. **Tinea pedis= athlete's foot (Moccasin & Interdigital & Vesicular & Ulcerative & Purpuric)**
5429. **One-hand-two-feet tinea syndrome**
5430. **Tinea profunda**
5431. **Tinea versicolor TVC =pityriasis versicolor PVC=Dermatomycosis furfuracea= Tinea flava=Tinea alba =Achromia parasitica**
5432. **Atrophic tinea versicolor TVC**
5433. **Erythrasmoid pityriasis versicolor**
5434. **Follicular Tinea versicolor TVC/Confetti-like pityriasis versicolor**
5435. **Tinea unguium**
5436. **Tumor necrosis factor receptor-associated periodic fever syndrome (TRAPS)= familiar Hibernian fever**
5437. **Bifid tongue**

5438. **Black hairy tongue = Lingua villosa nigra**
5439. **White hairy tongue**
5440. **Caviar tongue=Sublingual varicosities**
5441. **Scrotal Tongue=Fissured= plicated tongue=Furrowed**
5442. **Furred tongue**
5443. **Crenated tongue**
5444. **Pigmented Fungiform papilla of tongue**
5445. **Geographic Tongue (stomatitis areata migrans=Benign migratory glossitis= Annulus migrans)**
5446. **Smooth Tongue**
5447. **Topical steroid damaged face (TSDF)**
5448. **Toponymous contributions (see next)**
5449. ✓ **Acne mallorca**
5450. ✓ **Acroangiodermatitis of Mali**
5451. ✓ **African eye worm infection**
5452. ✓ **African Endemic Kaposi sarcoma**
5453. ✓ **African histoplasmosis**
5454. ✓ **African tick bite fever**
5455. ✓ **African trypanosomiasis**
5456. ✓ **Aleppo boil**
5457. ✓ **Ambras syndrome**
5458. ✓ **American hookworm infection**
5459. ✓ **American trypanosomiasis**
5460. ✓ **Andean sickness=white leprosy**
5461. ✓ **Balsam of Peru**
5462. ✓ **Biskra button**
5463. ✓ **Brazilian purpuric fever**
5464. ✓ **Bruton disease**
5465. ✓ **Burdwan fever**
5466. ✓ **Buruli ulcer**
5467. ✓ **Calabar swelling**

5468. ✓ Calcutta ulcer
5469. ✓ Congo red stain
5470. ✓ Coast of california calms neurofibromatosis
5471. ✓ Coast of maine mccune-albright syndrome
5472. ✓ Delhi boil
5473. ✓ Dum dum fever
5474. ✓ European blastomycosis
5475. ✓ Familial mediterranean fever
5476. ✓ Fort chaffee fever
5477. ✓ French disease for syphilis
5478. ✓ Gambian trypanosomiasis
5479. ✓ Haverhill fever
5480. ✓ Hibernian fever
5481. ✓ Indian filing
5482. ✓ Indian tick typhus
5483. ✓ Jaipur block
5484. ✓ Jodhpur technique
5485. ✓ Kandahar sore
5486. ✓ Lahore sore
5487. ✓ Lyme disease
5488. ✓ Madura foot
5489. ✓ Mal de maleda ppk
5490. ✓ Marseilles fever
5491. ✓ Mongolian spots
5492. ✓ Morbihan disease
5493. ✓ Nagashima palmoplantar keratoderma
5494. ✓ Naxos disease
5495. ✓ North america blastomycosis
5496. ✓ North carolina fever
5497. ✓ Norwegian scabies
5498. ✓ Ogna type epidermolysis bullosa simplex

5499. ✓ Ohio river valley fever
5500. ✓ Peruvian warts
5501. ✓ Pahvant valley fever
5502. ✓ Rocky mountain spotted fever
5503. ✓ Rose Bengal stain
5504. ✓ San joaquin valley fever
5505. ✓ Silk route disease
5506. ✓ South American blastomycosis
5507. ✓ Tokelau
5508. ✓ Tularemia
5509. ✓ Volhynia fever
5510. ✓ West nile fever
5511. ✓ Ebola virus disease
5512. Torch syndrome
5513. Torus mandibularis
5514. Torus palatine
5515. Touraine-Solente-Gole syndrome
5516. Toxic epidermal necrolysis TEN
5517. Toxic oil epidemic syndrome
5518. Toxic shock syndrome
5519. Toxocariasis
5520. Toxoplasmosis
5521. Transient abdominal telangiectasia of the newborn
5522. Transient acantholytic dermatosis
5523. Transient aquagenic wrinkling of palms
5524. Transient bullous dermolysis of the newborn
5525. Transient hypogammaglobulinemia of infancy
5526. Transient infantile lingual leukoplakia
5527. Transient lingual papillitis
5528. Transient localized hyperpigmentation newborn
5529. Transient neonatal cutis laxa
5530. Transient neonatal hair loss

5531. **Transient neonatal porphyrinaemia**
5532. **Transient neonatal pustular melanosis=Lentigines neonatorum**
5533. **Transient neonatal zinc deficiency**
5534. **Transient pigmentary lines of the newborn**
5535. **Transient telangiectatic purpura**
5536. **Transitory contractile dermatoses**
5537. **Transplantation related diseases**
5538. **Transporter associated with antigen processing TAP deficiency**
5539. **Traumatic ulcer**
5540. **Traumatic ulcerative granuloma**
5541. **Treacher Collin syndrome**
5542. **Tree man syndrome**
5543. **Trench fever**
5544. **Trench foot**
5545. **Manitoba- oculo- tricho- anal syndrome**
5546. **Trichiasis**
5547. **Trichinosis**
5548. **Tricho Entero Hepatic syndrome THES=phenotypic diarrhea**
5549. **Tricho oculo dermo vertebral syndrome**
5550. **Tricho retino dento digital syndrome**
5551. **Trichoadenoma**
5552. **Verrucous trichoadenoma**
5553. **Trichoadenoma of Nikolowski**
5554. **Trichobacteriosis pubis**
5555. **Trichoblastic carcinoma**
5556. **Trichoblastic carcinosarcoma**
5557. **Trichoblastic infundibular cyst TBIC**
5558. **Trichoblastoma**
5559. **Desmoplastic trichoblastoma**
5560. **Trichoclasis**
5561. **Trichodental syndrome**
5562. **Tricho-dento-osseous syndrome**
5563. **Trichodiscoma**

5564. **Trichodynia**
5565. **Trichodysplasia spinulosa**
5566. **Trichodysplasia‑amelogenesis imperfecta syndrome**
5567. **Trichoepithlioma**
5568. **Desmoplastic trichoepithelioma=sclerosing epithelial hamartoma**
5569. **Trichofolliculoma**
5570. **Trichogerminoma**
5571. **Tricholemmoma**
5572. **Desmoplastic tricholemmoma**
5573. **Malignant proliferating trichilemmal tumor**
5574. **Trichilemmal carcinoma**
5575. **Trichilemmal cyst**
5576. **Trichomadesis**
5577. **Trichomalacia**
5578. **Trichomatricoma**
5579. **Trichomegaly**
5580. **Trichomoniasis**
5581. **Trichomycosis axillaris**
5582. **Trichonodosis**
5583. **Tricho-odonto-onychial dysplasia**
5584. **Tricho-odonto-onycho dermal syndrome**
5585. **Tricho-onycho dental syndrome**
5586. **Trichopathophobia**
5587. **Trichopathy**
5588. **Trichophagia**
5589. **Trichophyton verrucosum**
5590. **Trichoptilosis**
5591. **Trichorrhexis invaginata**
5592. **Trichorrhexis nodosa**
5593. **Trichorhinophalangeal syndrome= Langer–Giedion**
5594. **Trichorhizophagia**
5595. **Trichorrhea**
5596. **Trichoschisis**

5597. **Trichoschisis/onychodystrophy**
5598. **Trichosporonosis**
5599. **Trichostasis spinulosa**
5600. **Trichothiodystrophy**
5601. **Trichotillomania (see next)= Alopecia Artefacta**
5602. ✓ **Trichoteiromania=Frictional alopecia**
5603. ✓ **Trichotemnomania**
5604. ✓ **Trichodaganomania**
5605. ✓ **Telogen mania**
5606. ✓ **Trichophobia**
5607. ✓ **Trichorrexomania**
5608. ✓ **Trichotillomania Incognito**
5609. ✓ **Trichobezoar**
5610. ✓ **Trichophytobezoar**
5611. **Trisomy 13 (patau syndrome)**
5612. **Tripe palms**
5613. **Trigeminal neuralgia**
5614. **Trigeminal trophic syndrome**
5615. **Traumatic ulcerative granuloma with stromal eosinophilia**
5616. **Trimethylaminuria**
5617. **Trombidiasis**
5618. **Trousseau's sign**
5619. **Trumpeter's wart**
5620. **African Trypanosomiasis = Sleeping sickness**
5621. **American Trypanosomiasis= Chagas disease**
5622. **Tuberous sclerosis complex TSC=(Bourneville disease & Epiloia & Angiofibroma & Shagreen patch & Ash-leaf macules &Koenen tumour & Periungual fibroma & Molluscum pendulum & Fibrous cephalic plaques & White freckles&Folliculocystic and collagenous hamartoma FCCH)**

5623. **Tuberculosis TB (see next)**
5624. ✓ **Gumma**
5625. ✓ **Lupus vulgaris=tuberculosis lupus (Plaque& Hypertrophic& Ulcerative&Vegetative)**
5626. ✓ **Acute military cutaneous tuberculosis**
5627. ✓ **Tuberculosis cutis orificialis**
5628. ✓ **Papulonecrotic tuberclid**
5629. ✓ **Rosacea-like tuberculid of Lewandowsky**
5630. ✓ **Lichen scrofulosorum=Tuberculosis cutis lichenoides**
5631. ✓ **Primary cutaneous tuberculosis complex=chancre**
5632. ✓ **Scrofuloderma=tuberculosis colliquativa cutis**
5633. ✓ **TB verrucous cutis=warty lupus**
5634. ✓ **BCGitis**
5635. ✓ **Tuberculous pseudomycetoma**
5636. ✓ **Tuberculous mastitis**
5637. **Mycobacterial spindle cell pseudotumor**
5638. **Mycobacterium abcess**
5639. **Mycobacterium avium -intracellulare complex**
5640. **Mycobacterium chelonae**
5641. **Mycobacterium fortuitum**
5642. **Mycobacterium gordonae disease**
5643. **Mycobacterium haemophilum**
5644. **Mycobacterium indicus pranii**
5645. **Mycobacterium kansasii**
5646. **Mycobacterium llatzerense**
5647. **Mycobacterium malmoense disease**
5648. **Mycobacterium marinum =fish tank granuloma=Swimming pool granuloma**
5649. **Mycobacterium scrofulaceum**
5650. **Mycobacterium szulgai disease**
5651. **Mycobacterium ulceran= Buruli ulcer**
5652. **Mycobacterium xenopi disease**

5653. **Tubular carcinoma**
5654. **Tufted angioma =Hypertrophic hemangioma=progressive capillary hemangioma=Angioblastoma of Nakagawa**
5655. **Tufted folliculitis**
5656. **Tularemia**
5657. **Tumour of the follicular infundibulum=infundibuloma; isthmicoma**
5658. **Tumoral melanosis**
5659. **Tungiasis**
5660. **Tunneling hair syndrome**
5661. **Turban tumor**
5662. **Turcot syndrome**
5663. **Turkey Gobbler neck /cervicomental**
5664. **Turner syndrome**
5665. **Twisted and rolled body hairs**
5666. **Tyloma**
5667. **Tylosis**
5668. **Type 1 Bare lymphocyte syndrome**
5669. **Typhoid &salmonellosis**
5670. **Epidemic (Louse -Borne) typhus**
5671. **Flying squirrel typhus**
5672. **Murine typhus=Endemic Typhus**
5673. **Scrub typhus= Tsutsugamushi disease= Mite-Borne typhus**
5674. **North Asian Tick Typhus (Siberian Tick Typhus)**
5675. **Queensland Tick Typhus**
5676. **Sporadic typhus=Brill–Zinsser disease**
5677. **Sylvatic Typhus**
5678. **Tyrosinemia**
5679. **Tyson's Glands**
5680. **Ulcer**
5681. **Ulcer in antiphospholipid syndrome**
5682. **Arterial Ulcer**
5683. **Chronic plantar Ulcer**
5684. **Decubitus Ulcer**

5685. **Pressure sores (bedsores)**
5686. **Diabetic ulcer**
5687. **Genital Ulcer**
5688. **Ulcerated gouty tophi**
5689. **Ulcer due to Hematologic Disorders**
5690. **Ischemic ulcer**
5691. **Lipschutz ulcer / Lipschütz**
5692. **Marjolin ulcer**
5693. **Meleney ulcer**
5694. **Martorel ulcer**
5695. **Neuropathic ulcer**
5696. **Ulcer in rheumatoid arthritis**
5697. **Ulcer in sickel cell anemia**
5698. **Tropical Ulcer=tropical phagedaena**
5699. **Ulcer in Thalassemia**
5700. **Ulcer in vasculitis**
5701. **Venous ulcer**
5702. **Ulerythema ophryogenes =keratosis pilaris atrophicans faciei**
5703. **Ulerythema sycosiforme**
5704. **Ulnar–mammary syndrome**
5705. **Umblical granuloma**
5706. **Uncombable hair syndrome UHS**
5707. **Uncombable hair, retinal pigmentary dystrophy, dental anomalies and brachydactyly (Bork syndrome)**
5708. **Undifferentiated connective tissue disease(UCTD)**
5709. **Unilateral laterothoracic exanthema=asymmetric periflexural exanthem of childhood**
5710. **Unilateral naevoid telangiectasia**
5711. **Universal angiomatosis**
5712. **Universal Lightening of Hair**
5713. **Uncombable hair syndrome**
5714. **Unverricht–Lundborg Disease**
5715. **Upper dermal elastolysis**

5716. **Upshaw- schulman syndrome**
5717. **Urachus cyst**
5718. **Urbach-wiethe disease**
5719. **Uremic follicular hyperkeratosis**
5720. **Uremic frost**
5721. **Urticaria= Hives (Acute & Chronic & Recurrent)**
5722. **Adrenergic urticaria**
5723. **Aquagenic urticaria**
5724. **Cholinergic urticaria**
5725. **Cold urticaria**
5726. **Essential acquired cold urticaria**
5727. **Primary cold contact urticaria**
5728. **Systemic cold urticaria**
5729. **Reflex cold urticaria**
5730. **Contact urticaria**
5731. **Exercise-induced urticaria**
5732. **Exercise induced anaphylaxis**
5733. **Galvanic Urticaria**
5734. **Heat urticaria**
5735. **Papular urticaria=Insect bite hypersensitivity**
5736. **Pressure urticaria**
5737. **Psychogenic Urticaria**
5738. **Recall urticaria**
5739. **Solar urticaria**
5740. **physical type urticaria**
5741. **Vibratory angioedema**
5742. **Yellow Urticaria**
5743. **Urticarial dermatitis**
5744. **Urticaria-like follicular mucinosis**
5745. **Urticaria multiforme**
5746. **Urticaria pigmentosa**
5747. **Urticarial vasculitis**
5748. **Uv-sensitive syndrome**

5749. **Vaccinia**
5750. **Generalized vaccinia**
5751. **Progressive vaccinia (vaccinia necrosum,vaccinia gangrenosum)**
5752. **Vaccination-associated morphea**
5753. **Vagabond's disease**
5754. **Valley fever**
5755. **Van Allen-Myhre syndrome**
5756. **Van der Woude syndrome**
5757. **Van Lohuizen syndrome**
5758. **Varicella& Varicella gangrenosum**
5759. **Sublingual varices**
5760. **Vascular malformations (see next)**
5761. **✓Capillary malformation**
5762. **✓Lymphatic malformation**
5763. **✓Mixed capillary-venous-lymphatic malformation**
5764. **✓Arteriovenous malformation**
5765. **✓Capillary malformation-arteriovenous malformation(CM-AVM)**
5766. **Vascular twin spotting (vts)**
5767. **Vasculitis: drug induced**
5768. **Hypersensitivity Vasculitis**
5769. **Leukocytoclastic Vasculitis LCV**
5770. **Vasculitis: septic type**
5771. **Single organ vasculitis**
5772. **Localized chronic fibrosing vasculitis**
5773. **Vasculitis in Systemic lupus erythematosus**
5774. **Vasculitis in Rheumatoid arthritis**
5775. **Vasculitis: Small vessel type**
5776. **Vasculitis:Medium vessel type**
5777. **Vasculitis:Large vessel type**
5778. **Vein graft site dermatitis**
5779. **Vellus hair cyst**
5780. **Velocardiofacial syndrome**

5781. Venomous Fish= Stinging fish
5782. Venous lake=Senile Hemangioma
5783. Venous malformation
5784. Cephalic venous malformation
5785. Verrucous venous malformation =verrucous haemangioma
5786. Vernix caseousa
5787. Verruca vulgaris (VV)
5788. Verrucal keratosis
5789. BRAF inhibitor-associated verrucous keratosis (BAVK)
5790. Verrucous carcinoma
5791. Verrucous cysts
5792. Verrucous hemangioma=Verrucous Venulocapillary Malformation
5793. Verrucous leukoplakia
5794. Verrucous perforating collagenoma (collagenome perforant verruciformé)
5795. Verrucous skin lesions in diabetic neuropathy (VSLDN)
5796. Verrucous xanthoma= verruciform xanthomas
5797. Verruga peruana =peruvian warts=Oroya fever =Carrion disease= Bartonellosis
5798. Vestibular papillomatosis
5799. VEXAS syndrome(Vacuoles,E1 enzyme,X-linked,autoin6ammatory,somatic)
5800. Vibration white finger
5801. Vibrio bacteria related infections
5802. Vici syndrome
5803. Villar's nodule =umbilical endometriosis
5804. Viral exanthema
5805. Non-specific viral eruptions
5806. Viral hemorrhagic fevers
5807. Old-World Hemorrhagic Fevers (Arenaviruses :Lassa virus&Filoviruses :Marburg and Ebola& Bunyaviruses& Hantaviruses)
5808. New-World Hemorrhagic Fevers(Hantaviruses&Arenaviruses)
5809. Vitamin A (retinol) deficiency

5810. **Vitamin B deficiency(B1= Thiamine,B2= Riboflavin,B3= Niacin,B5= pantothenic acid,B6= Pyridoxine,B7 =biotin,B9= Folic acid,B12= Cobalamin)**
5811. **Vitamin C (Ascorbic acid) deficiency**
5812. **Vitamin D (calcitriol) deficiency**
5813. **Vitamin D-dependent rickets with alopecia**
5814. **Vitamin k deficiency= phytonadione deficiency**
5815. **Vitiligo**
5816. **Inflammatory vitiligo**
5817. **Trichrome vitiligo**
5818. **Quadrichrome vitiligo**
5819. **Pentachrome vitiligo**
5820. **Hypochromic vitiligo (HV)**
5821. **Blue vitiligo**
5822. **Burn vitiligo**
5823. **Papulosquamous vitiligo**
5824. **Vitiligo en confetti**
5825. **vitiligo-like acquired dermal elastolysis**
5826. **Vogt–Koyanagi–Harada syndrome**
5827. **Volkmann's contracture**
5828. **Von Hippel-Lindau syndrome**
5829. **Von Recklinghausen's disease**
5830. **Von Zumbusch : pustular psoriasis**
5831. **VRESS :virus reactivation with eosinophilia and systemic symptoms**
5832. **Massive Vulval Edema**
5833. **Vulvar intraepithelial neoplasia VIN**
5834. **Vulvar melanosis VM**
5835. **Vulvitis granulomatosis**
5836. **Vulvovaginal gingival syndrome= Hewitt-pelisse**
5837. **Vulvodynia**
5838. **Walbaum –dehane–schlemmer syndrome**
5839. **Waardenburg syndrome (4types)**
5840. **Hypergammaglobulinemic purpura of waldenström**
5841. **Walking dandruff= Cheyletiellosis**

5842. **Warburg-Cinotti syndrome**
5843. **Warfarin induced diseases**
5844. **Warfarin associated venous limb gangrene**
5845. **Warfarin necrosis**
5846. **Wart (see next)**
5847. **✓Common wart = verruca vulgaris**
5848. **✓ Plane wart = verruca plana**
5849. **✓ Plantar wart**
5850. **✓Digitate wart**
5851. **✓filiform wart**
5852. **✓Butcher's wart**
5853. **✓Genital wart =Condylomata Acuminata**
5854. **✓Doughnut wart**
5855. **✓Mirmecia**
5856. **✓Anal wart**
5857. **✓Kissing wart**
5858. **✓Mucous membrane wart**
5859. **✓Punctate wart**
5860. **✓Periungual and subungual wart**
5861. **✓ Cystic wart**
5862. **Warty dyskeratoma**
5863. **Warthin tumor (papillary cystadenoma lymphomatosum)**
5864. **Wattle**
5865. **Watson syndrome**
5866. **Waxy keratosis= kerinokeratosis papulosa =waxy keratosis of childhood**
5867. **Weathering nodule**
5868. **Weaver's syndrome**
5869. **Wiedemann–Rautenstrauch syndrome= Neonatal progeroid syndrome**
5870. **Wegener's granulomatosis**
5871. **Weightlifter's nodule**
5872. **Weill–Marchesani syndrome**

5873. **Wells syndrome =Eosinophilic cellulitis (plaque & annular & papulonodular & papulovesicular & bullous & linear & urticarial like & FDE like)**
5874. **Wende–bauckus/pegum syndrome**
5875. **Werewolf syndrome**
5876. **Werlhof's disease**
5877. **Wermer's syndrome**
5878. **Werner's syndrome**
5879. **West nile virus infection**
5880. **Weyer acrofacial dysostosis (acrodental dysostosis of weyer, curry–hall syndrome)**
5881. **Whipple disease (subcutaneous)**
5882. **Whitlow**
5883. **Whim syndrome**
5884. **Whisker Hairs**
5885. **Whitmore's disease**
5886. **White fibrous papulosis of neck**
5887. **White forelock with osteopathia striata**
5888. **White forelock with multiple malformations**
5889. **White sponge nevus of Cannon = familial white folded gingivostomatitis**
5890. **Wickham's striae**
5891. **Wiedemann-rautenstrauch syndrome**
5892. **WILD syndrome (warts, immunodeficiency, lymphedema, anogenital dysplasia)**
5893. **Williams elfin facies syndrome**
5894. **Williams–Beuren syndrome**
5895. **Winchester's syndrome**
5896. **Windmill-Vane-Hand syndrome**
5897. **Witkop–Brearley–Gentry syndrome= Hypoplastic enamel–onycholysis–hypohidrosis**
5898. **Witkop–von sallmann syndrome**
5899. **Wilson disease**
5900. **Wiskott–Aldrich syndrome**
5901. **Wnt 10 disorders**

5902. **Wolf-man syndrome**
5903. **Woolf syndrome**
5904. **Hereditary woolly hair HWH**
5905. **Woolly hair nevus**
5906. **Woolly hair syndrome**
5907. **Woolly hair-skin fragility syndrome (WHSF)**
5908. **Woolly hair, hypotrichosis, everted lower lip and outstanding ears (salamon syndrome)**
5909. **Autosomal recessive Woolly hair/hypotrichosis**
5910. **Wolf–Hirschhorn syndrome**
5911. **Wolf 's postherpetic isotopic response**
5912. **Wohlfahrtiimonas chitiniclastica**
5913. **Wong type dermatomyositis**
5914. **Woronoff's ring**
5915. **Woringer kolopp type pagetoid reticularis**
5916. **Wound**
5917. **Wrinkles**
5918. **Wrinkling skin syndrome**
5919. **Wrinkly skin syndrome**
5920. **Wyburn–mason syndrome**
5921. **Xanthelasma palpebrarum**
5922. **Xanthoma (tuberous & eruptive & tendenous & tuberoeruptive & plane)**
5923. **Cerebrotendinous Xanthomatosis**
5924. **Xanthotrichia**
5925. **Xanthomatous giant cell sarcoma**
5926. **Xeroderma pigmentosum (8 genotypes)(A,B,C,D,E,F,G,V)**
5927. **Pigmented xerodermoid**
5928. **Xeroderma pigmentosum/Cockayne syndrome complex**
5929. **Xeroderma, talipes, and enamel defect**
5930. **Xerosis cutis**
5931. **X-linked agammaglobulinemia**
5932. **X-linked hypodontia**
5933. **X-linked lymphoproliferative syndrome (duncan disease)**

5934. **X-linked reticulate pigmentary disorder=partington syndrome= Familial cutaneous amyloidosis**
5935. **X-linked severe combined immunodeficiency (SCID)**
5936. **Xmen (x-linked immunodeficiency with magnesium defect, ebv infection, and neoplasia)**
5937. **X-shaped bluish hyperpigmentation in hypohidrotic ectodermal dysplasia**
5938. **Yao syndrome**
5939. **Yaws=pian, frambesia, bouba**
5940. **Yellow fever**
5941. **Yemenite deaf-blind hypopigmentation syndrom=Warburg-Thomsen syndrome**
5942. **Yersenia pestis related diseases**
5943. **Yunis-varon syndrome**
5944. **Zanier–Roubicek syndrome**
5945. **Zellweger syndrome**
5946. **Zirconium granuloma**
5947. **Zika fever**
5948. **Congenital Zika syndrome**
5949. **Zimmerman –Laband syndrome**
5950. **Zinser–Engmann–Cole syndrome**
5951. **Ziprkowski–Margolis syndrome (Albinism–Deafness syndrome)**
5952. **Zirconium granuloma**
5953. **Zlotogora–ogur syndrome**
5954. **Zoon balanitis= plasma cell balanitis= Balanitis circumscripta plasmacellularis**
5955. **Zoon vulvitis**
5956. **Zosteriform dermatoses**
5957. **Zosteriform metastasis**
5958. **Zosteriform Venous malformations =Unilateral segmental Cavernous Hemangiomatosis**
5959. **Zouboulis syndrome**
5960. **Zunich–kaye syndrome=Zunich neuroectodermal syndrome**
5961. **Zygomycosis =Mucormycosis=Phycomycosis**

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Dermatology diseases index

2025 · Hany salah soliman

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... nevus (CMN) 3676. Bathing trunk nevus =Giant pigmented nevus 3677. Congenital panfollicular nevus 3678. Connective tissue...

netic factors. Now in the early twenty-first century, a concerted effort between bench researchers, clinicians, and pathologists is needed to further our understanding about this diverse, intriguing group of conditions.

11.1 Approach to the Patient with Cicatricial Alopecia

Prompt diagnosis and treatment of active disease to halt further hair loss is the goal. Clinical diagnosis of primary cicatricial alopecia should be confirmed by histopathology. A single 4-mm punch biopsy sample is recommended, taking care to choose a biopsy site with clinically representative active disease which is usually at the advancing, hair-bearing edge of an area of alopecia. Symptoms, inflammatory stigmata, and a positive pull test are helpful signs. Transverse sectioning of the biopsy specimen is conventionally advocated to ensure that all follicles are assessed at multiple planes; however, loss of critical diagnostic features in the epidermis, superficial dermis, and panniculus with this mode can impair interpretation [49, 69]. Obtaining a second specimen for vertical sectioning can improve diagnostic yield, but may still not allow distinction among predominantly lymphocytic or neutrophilic inflammation [48, 111]. The authors use this dual approach when faced with clinically puzzling cases and when discoid lupus erythematosus is considered. The specimen to be sectioned vertically is bisected, with half sent for hematoxylin-eosin staining and the other for direct immunofluorescence. Elastin stains can be used in more advanced disease to help distinguish between the more common lymphocytic predominant disorders (which often overlap both clinically and histopathologically) based on different patterns of scar formation [47].

Once the diagnosis is established, active disease must be treated immediately to thwart further hair loss. Although hampered in large part by the lack of formal, controlled studies, there are ample empiric data in most conditions to devise a tiered treatment plan. Flexibility is key. In some, the hair loss continues despite these measures, and cosmetic aids and supportive care are what are needed most.

Table 11.1 Working classification of primary cicatricial alopecias based on predominant inflammatory cell involved[a]

Lymphocytic	**Lichen planopilaris**
	– **Sparse, curly, and fair hair**
	– **Classic variant**
	– **Frontal fibrosing alopecia**
	– **Piccardi–Lassueur–Graham Little syndrome**
	– **Fibrosing alopecia in a pattern distribution**
	Pseudopelade of Brocq
	Central centrifugal cicatricial alopecia
	Alopecia mucinosa
	Keratosis pilaris spinulosa decalvans/ Folliculitis spinulosa decalvans
	Discoid lupus erythematosus
Neutrophilic	**Folliculitis decalvans**
	Perifolliculitis capitis abscedens et suffodiens
Mixed	**Acne keloidalis**
	Acne necrotica
	Erosive pustular dermatosis of the scalp

[a] Revised from NAHRS classification scheme, *J Am Acad Dermatol* 2003;48:103-10 [123]. Entities in *italics* may not be appropriately classified, distinct, or clearly primary in nature. See text for details.

11.2 Classification

A standardized classification scheme was adopted by the North American Hair Research Society in 2001 as a fulcrum for further discussion, debate, and research [123]. It is based on the primary inflammatory cell involved in active disease (lymphocytic, neutrophilic, or mixed) (Table 11.1). Modification is expected as advances occur. The chapter will address the entities listed in Table 11.1 in the order in which they appear.

11.3 Lymphocytic Disorders

11.3.1 Lichen Planopilaris

Lichen planopilaris is a follicular form of lichen planus. Three clinical variants result in cicatricial alopecia: (1) classic lichen planopilaris; (2) frontal fibrosing alopecia; and (3) Piccardi-Lassueur-Graham Little syndrome.

Table 12.1 Secondary cicatricial (permanent) alopecias, a classification based on etiology *(see next page)*

Secondary cicatricial (permanent) alopecias			
1. Genodermatoses and developmental defects with permanent alopecia (excluding congenital hypotrichoses and atrichias)	Ectodermal dysplasias	*4. Inflammatory dermatoses*	
	Aplasia cutis congenita	*Pseudo-cicatricial*	Pyoderma gangraenosum
	Incontinentia pigmenti		Graft-versus-host disease
	Porokeratosis mibelli		*Sclerosing*
	Ichthyosis		Morphea
	Hereditary epidermolysis bullosa		Scleroderma en coup de sabre and Parry-Romberg syndrome
	Meningocele		Lichen sclerosus et atrophicus
	Hamartoma		*Bullous*
	Organoid nevi (sebaceous, epidermal)		Cicatricial pemphigoid
	Vascular malformations		Porphyria cutanea tarda
	Darier's disease		Acquired Epidermolysis Bullosa
	Fibrodysplasia	**Displacement**	*Granulomatous*
2. Physical and chemical injury	Mechanical trauma and pressure		Sarcoidosis
True cicatricial	Scratching		Granuloma anulare
	Burns		Necrobiosis lipoidica (including Miescher's granulomatosis)
	Freezing	**5. Drugs**	
	Chemical injury	**6. Neoplasms**	
	Insect bites	**Displacement**	*Infiltration*
Pseudo-cicatricial	Radiation		Lymphoproliferative disorders
3. Infections			Mastocytosis
True cicatricial	*Bacterial*		*Benign solid neoplasms*
	Leprosy		Cysts
	Tertiary syphilis		Vascular tumors
	Tuberculosis-lupus vulgaris		Adnexal tumors
	Viral		Plasmocytoma
	Zoster		*Malignant solid tumors*
	Varicella		Angiosarcoma
	Tinea capitis		Dermatofibroscarcoma protuberans
	Kerion		Malignant fibrous histiocytoma
	Favus		Melanoma
	Protozoic		Squamous cell carcinoma
	Leishmania		Basal cell carcinoma
4. Inflammatory dermatoses			Metastasis (alopecia neoplastica)
Pseudo-cicatricial	Psoriasis (rarely)		Lymphoma
	Pityriasis amiantacea		
	Arteriitis temporalis		

TABLE 1 | Entities comprising the proposed frontal fibrosing alopecia syndrome (FFA Syndrome).

FFA variants
A. Classic FFA • Grades 1-5 (severity) • Common patterns ◦ Linear (type 1), diffuse (type 2) and pseudo-fringe (type 3)
B. Uncommon/Atypical FFA variants ◦ Androgenetic Alopecia-like ◦ Cockade-like pattern ◦ Clown Alopecia pattern ◦ Occipital patterns (ophiasis-like, lateral, patchy, pseudo-fringe) ◦ Upsilon pattern ◦ Pustular
C. FFA with associated cutaneous or nail changes • FFA associated with classic scalp lichen planopilaris • FFA associated with fibrosing alopecia in a pattern distribution • FFA associated with Graham-Little-Piccardi-Lassueur Syndrome • FFA associated with facial papules • FFA associated with nuchal papules and ectatic veins • FFA associated with severe cutaneous atrophy/depression of the frontal veins • FFA associated with rosacea • FFA associated with follicular red dots (glabella/ forehead, eyebrows, hips, cheeks, chest) • FFA associated with androgenetic alopecia • FFA associated with irritant/allergic contact dermatitis • FFA associated with psoriasis/seborrheic dermatitis • FFA associated with excessive facial sweating • FFA associated with lichen planus pigmentosus • FFA associated with reversal of canities • FFA associated with cutaneous lichen planus/ FFA associated with nail lichen planus • FFA associated with atopy
D. FFA initially presenting as isolated non-scalp hair loss presentations • FFA presenting with isolated eyebrow loss (no scalp hair loss) • FFA presenting with rapid loss of body hair (no scalp hair loss)
E. FFA with mucosal changes • FFA with lichen sclerosus • FFA with genital lichen planus • FFA with meibomian gland dysfunction • FFA with oral lichen planus
F. FFA with comorbid autoimmune diseases • FFA with (i) alopecia areata, (ii) cutaneous/systemic lupus, (iii) vitiligo, (iv) Sjogren's syndrome, (v) rheumatoid arthritis, (vi) inflammatory bowel disease, (vii) others
G. FFA with comorbid endocrine diseases • FFA with thyroid disorders • FFA with androgen excess/ FFA with androgen deficiency • FFA associated with vitamin D deficiency • FFA associated with metabolic dysfunction/dyslipidemia • FFA post-hysterectomy/FFA with ovarian insufficiency
H. FFA with early age of onset • FFA with premenopausal onset • FFA with premenopausal onset PLUS ovarian insufficiency • FFA with adolescent or pre-pubertal onset
I. FFA in special groups • FFA in males • FFA in skin of color

(Continues)

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TABLE 1 | (Continued)

FFA variants
J. FFA with family history • Familial FFA
K. Medication-induced FFA • FFA associated with use of medications such as: tamoxifen, raloxifene, checkpoint inhibitors, prostate cancer medications, estrogens
L. Trauma-induced FFA • FFA developing post: (i) hair transplant, (ii) facelift surgery, or (iii) laser ablative resurfacing

TABLE 2 | Example of 2018 Proposed Diagnostic Criteria for FFA and Limitations in LPPAI.

Part 1: (2018) Vañó-Galván et al. Frontal Fibrosing Alopecia Diagnostic Criteria. Diagnosis requires 2 major criteria or 1 major criterion and 2 minor criteria. (Modified from Vañó-Galván et al. [2]).

Major criteria

1. Clinical appearance of scarring alopecia of the frontal, frontotemporal or temporal scalp, in the absence of follicular keratotic papules on the body
2. Diffuse bilateral eyebrow loss

Minor criteria

3. Typical trichoscopic features of the disease: perifollicular erythema and/or follicular hyperkeratosis
4. Histopathologic features consistent with LPP/FFA
5. Hair loss at sites other than the frontal hairline: occipital area, facial hair, sideburns, or body hair
6. Noninflammatory facial papules

Part 2: Example of limitation of using percent changes in Lichen Planopilaris Activity Index (LPPAI) to evaluate FFA treatment efficacy.

Medication	LPPAI before treatment	LPPAI after treatment	Regrowth in eyebrows?	Regrowth in hairline?	Percent reduction in LPPAI	Most effective treatment
A	3.8	1.3	No	No	65.8%	
B	3.8	1.3	Yes	Yes	65.8%	B

Abbreviations: FFA, frontal fibrosing alopecia; LPP, Lichen planopilaris; LPPAI, Lichen Planopilaris Activity Index.

4 | Recognizing FFA as A Syndrome Would Open Up New Dialog Among Clinicians and Researchers

Much disagreement exists in our community regarding many aspects of FFA diagnosis and treatment. For example, we disagree on how best to treat FFA. A recent article by Vañó-Galván et al. proposing that dutasteride might best be viewed as a first-line treatment for FFA [3] was quickly followed by an opposing article by Holmes et al. [4] outlining why dutasteride should not be considered a first-line option.

Our current narrow view of FFA as a single disease entity only adds to the challenges of studying this complex condition. If we were more open to recognizing FFA as a syndrome with extremely varied clinical presentations, we might be more open to the possibility that different treatments might work better in different presentations. A new focus on FFA as a syndrome might open us all to consider the possibility that there might be more than one first-line treatment option for various presentations.

For example, it's difficult to argue against the use of isotretinoin as a first-line treatment for FFA associated with prominent facial papules. Isotretinoin, however, might not be an appropriate option for a patient with dry eyes secondary to severe meibomian gland dysfunction or Sjögren's syndrome. Similarly, dutasteride might be considered a reasonable first-line option for many FFA patients but is not an ideal option for a patient with low libido associated with severe androgen deficiency.

5 | Viewing FFA as a Syndrome Requires Us to Redefine What Constitutes a Treatment Success

Our current tunnel view of FFA as a single disease entity influences many practitioners to think first and foremost about what is happening to the hairline and leads many to ignore the 30 or more signs and symptoms associated with this syndrome (Table 1). A new focus on FFA as a syndrome would bring needed attention to these issues. This is important not only in diagnosis but also as we consider how best to define treatment success.

In my view, we need to define better what constitutes a successful treatment outcome. The current methods for evaluating treatment success have limitations—and often rely on tools that specifically assess disease activity or severity rather than assess

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Table 36.7: Classification of idiopathic inflammatory dermatomyopathies

Dermatomyositis (DM)

Adult onset

- Classic DM
  - Classic DM alone
  - Classic DM with malignancy
  - Classic DM as a part of an overlap connective tissue disorder
- Clinically amyopathic DM (C-ADM)
  - Amyopathic DM (ADM)
  - Hypomyopathic DM

Juvenile onset

- Classic DM
- Clinically amyopathic DM (C-ADM)
- Amyopathic DM (ADM)
- Hypomyopathic DM

Polymyositis (PM)

PM alone
PM as a part of an overlap connective tissue disorder
PM associated with malignancy

Inclusion body myositis

Other clinical pathologic subgroups of myositis

Focal myositis
Orbital myositis
Proliferative myositis
Eosinophilic myositis
Granulomatous myositis

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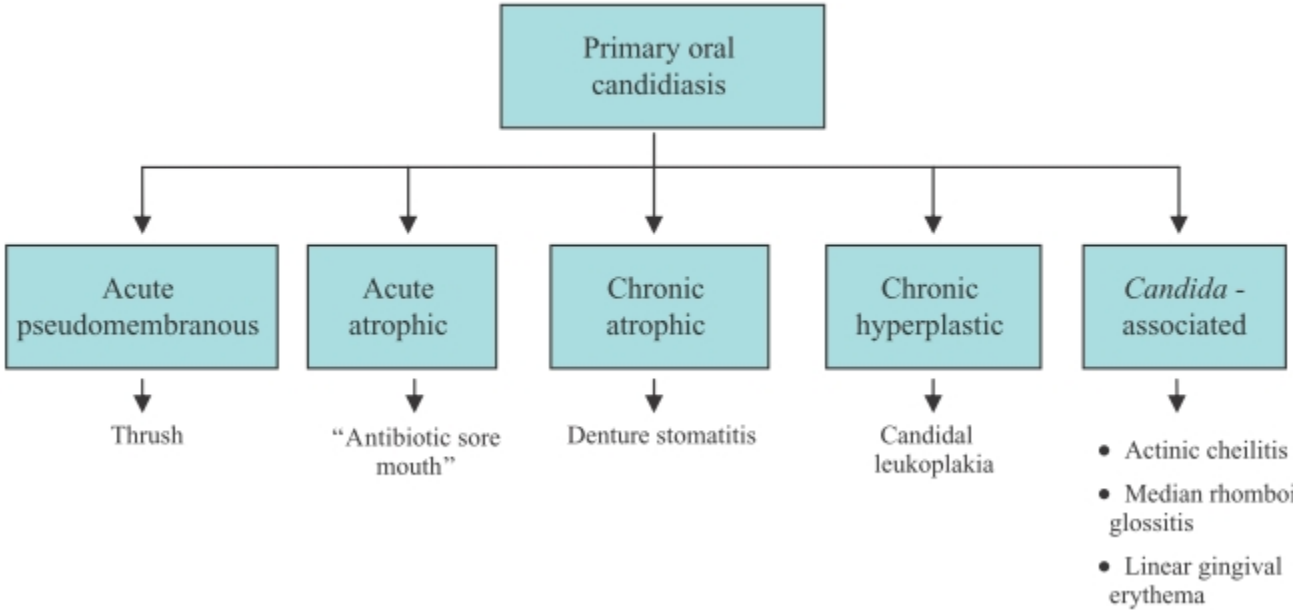

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Table 3. Classification of oral candidiasis (16)

Primary oral candidiasis	Secondary oral candidiasis
Acute forms Pseudomembranous Erythematous	Oral manifestations of systemic mucocutaneous candidiasis as a result of diseases such as a thymic aplasia and candidiasis endocrinopathy syndrome
Chronic forms Hyperplastic Nodular Plaquelike Erythematous	
Candida-associated lesions Denture stomatitis Angular cheilitis Median rhomboid glossitis Linear gingival erythema	

latter has now been recovered from many superficial and systemic lesions under various disease conditions.

Classification of oral *Candida* infections is fraught with difficulties due to the varied manifestations of the disease. It is generally accepted

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Table 1. Classification schemes for oral candidosis

Early Classification	Revised* (1990)	Proposed** (2005)
Acute pseudomembranous	Acute pseudomembranous	pseudomembranous
Acute atrophic	Acute erythematous	erythematous
Chronic hyperplastic	Chronic nodular Chronic plaque-like	hyperplastic
Chronic atrophic (Denture stomatitis)	Chronic erythematous (Denture-associated candidosis)	erythematous (Denture-induced stomatitis)
–	Chronic pseudomembranous	pseudomembranous
Angular cheilitis	Candida-associated cheilitis	Angular cheilitis
Median rhomboid glossitis	–	Median rhomboid glossitis

*Samarnayake LP, Yaacob HB. Classification of oral candidosis. In: Samaranayake LP, MacFarlane TW. Oral Candidosis. London: Wright, 1990:124-132.

**The proposed classification removes the terms 'acute' and 'chronic' as the temporal aspects of the clinical presentation have little bearing on the causality or treatment of the condition.

Table 2. Summary of a new classification for oral candidiasis

Primary	Secondary
Acute Pseudomembranous and erythematous mucocutaneous	Mucocutaneous presence as a result of systemic disease
Chronic Pseudomembranous, erythematous, hyperplastic, nodular and plaque-like	
***Candida*-associated** Denture stomatitis, angular chelitis and median rhomboid glottitis	
Keratinized primary lesion superinfected with *Candida* Leukoplakia, lichen planus and lupus erythematosus	

Sources: Axéll et al. (7) and Samaranyake & Yaacob (166)

SUBTYPES OF PRIMARY AND DRUG-INDUCED THROMBOTIC MICROANGIOPATHY (TMA)					
Subtype of TMA	**Etiology**	**Pathomechanism**	**Onset**	**Clinical features**	**Treatment**
Inherited					
ADAMTS13 deficiency-mediated [thrombotic thrombocytopenic purpura]	Homozygous or heterozygous mutations in *ADAMTS13*	ADAMTS13 is a vWf-cleaving protease and its dysfunction leads to vascular occlusion from ultra-large vWF multimers	Childhood, occasionally adulthood	Recurrent TMA; neurologic or other organ (but rarely severe renal) injury *Petechiae*	Plasma infusion, plasma exchange
Complement-mediated (90% of cases inherited)	Mutations in genes that encode components of the alternate complement pathway, e.g. H, I, B, C3, CD46	Uncontrolled activation of the alternative complement pathway	Often childhood, sometimes adulthood	TMA; renal injury common	Anti-complement therapy (e.g. eculizumab), plasma exchange or infusion
Metabolism-mediated	Mutations in *MMACHC* lead to AR disorder methylmalonic aciduria and homocystinuria, CblC type	Metabolic abnormalities trigger platelet activation, formation of reactive oxygen species, endothelial dysfunction, ↑tissue factor, and coagulation	Infancy (before 1 year of age)	Developmental delay, hypotonia; pulmonary artery hypertension; chronic kidney disease with hypertension and proteinuria	B12, betaine, folinic acid
Coagulation-mediated	Mutations in *THBD*, *PLG*, and *DGKE* which encode thrombomodulin, plasminogen, and diacylglycerol kinase epsilon (DGKE), respectively	DGKE dysfunction leads to upregulation of prothrombotic factors and downregulation of VEGF receptor	Usually infancy (before 1 year of age)	Acute kidney injury	Plasma infusion
Acquired					
ADAMTS13 deficiency-mediated [thrombotic thrombocytopenic purpura]	Autoantibodies lead to inhibition of ADAMTS13 activity (to <10% of normal)	Decreased activity of ADAMTS13 leads to vascular occlusion from ultra-large vWF multimers (see above)	Adulthood (usually 20–50 years of age) > childhood More common in women and blacks	TMA with thrombocytopenia; GI symptoms, focal neurologic defects, transient renal injury *Petechiae*	Plasma exchange (improves survival from 10% to ~80% but must be instituted early)
Shiga toxin (ST)-mediated [hemolytic uremic syndrome]	Enteric infection with a toxin-secreting strain of either *Escherichia coli* (especially O157:H7) or *Shigella dysenteriae*	Toxin binds to Gb3 (CD77, ceramide trihexoside) on endothelial, renal epithelial, and mesangial cells, with binding leading to apoptosis Toxin also prothrombotic	Early childhood (median age = 2 years; 3% mortality) > adulthood (more severe)	Infectious enterocolitis As diarrhea resolves, thrombocytopenia and renal disease develop	Supportive care including aggressive hydration, dialysis often required
Drug-mediated, immune type	Most commonly quinine, quetiapine, gemcitabine; also interferon-β (more likely than toxic)	With quinine, drug-dependent antibodies	Adulthood (drug exposure more likely)	Sudden onset of severe systemic symptoms; anuric renal injury	Discontinuation of drug, supportive care
Drug-mediated, toxic/dose-related	Multiple drugs, including cyclosporine, tacrolimus, VEGF inhibitors	Multiple mechanisms	Adulthood (drug exposure more likely)	Gradual onset of renal failure	Discontinuation of drug, supportive care
Complement-mediated (10% of cases acquired)	Antibodies inhibit complement factor H activity	Reduced inhibition of alternate complement pathway factors (i.e. enhanced complement activation) leads to vessel damage	Childhood or adulthood	Acute kidney injury	Plasma exchange, immunosuppression, anti-complement therapy (e.g. eculizumab)

Table 23.4 Subtypes of primary and drug-induced thrombotic microangiopathy (TMA). Disease may remain subclinical unless triggered by factors such as pregnancy, autoimmune connective tissue disease, systemic infections, hematopoietic stem cell transplant, malignant hypertension or drugs; this is referred to as secondary TMA. AR, autosomal recessive; VEGF, vascular endothelial growth factor; vWf, von Willibrand factor.

Interventions to correct the underlying PAD, such as femoral artery angioplasty or femoral popliteal bypass surgery, lead to rapid resolution. Less often, lesions resolve without therapy[65].

Hematologic Disorders

Anemia is a major and often underrecognized cause for delayed wound healing. It prevents proper tissue oxygenation. In addition, some forms of anemia are associated with a marked tendency for ulcer formation, such as the hemoglobinopathies (Fig. 105.20B)[66]. Hematologic malignancies are also associated with ulcerative processes including pyoderma gangrenosum, vasculitis, and cryoglobulinemia (particularly type I). Clotting abnormalities can play a direct role in the formation of ulcers (e.g. antiphospholipid antibody syndrome)[67,68], as well as serving as a predisposing factor for more common types of cutaneous ulcerations such as venous ulcers[69,70]. Acute massive venous thrombosis, also known as phlegmasia cerulea dolens, can lead to massive edema in the legs, with subsequent ischemia and ulceration due to venous limb gangrene[71]. Table 105.9 summarizes the recommended evaluation when thrombophilia is suspected.

EVALUATION FOR THROMBOPHILIA

Disorder	%*	Screening tests	Potential confounding conditions
Inherited			
Factor V Leiden	5	Activated protein C (APC) resistance†	Warfarin, heparin, OCP, pregnancy, ↑ factor VIII level, lupus anticoagulant†
	5	Factor V Leiden mutation (AD) [reflex test if APC detected]	–
Prothrombin G20210A	2	Prothrombin G20210A mutation (AD)	–
Protein C deficiency	0.3	↓ Protein C activity (AD)	**Warfarin**, OCP, pregnancy, liver disease, ↑ factor VIII level, lupus anticoagulant, acute thrombosis
Protein S deficiency	0.05	↓ Free protein S antigen level and/or activity (AD)	**Warfarin**, OCP, pregnancy, liver disease, ↑ factor VIII level, lupus anticoagulant, acute thrombosis
Antithrombin deficiency	0.1	↓ Antithrombin activity (AD)	**Heparin**, liver disease, acute thrombosis
Hyperhomocysteinemia	>5	↑ Homocysteine level (may have homozygous *MTHFR* C677T mutation or be compound heterozygote for *MTHFR* C677T/A1298C > heterozygous *CBS* mutation)	Deficient folate, B_{12} or B_6; older age, smoking
Excess factor VIII/von Willebrand factor	10	↑ Factor VIII level	Acute phase response, OCP, pregnancy, old age
Excess fibrinogen	ND	↑ Fibrinogen level	Acute phase response, pregnancy, smoking, older age
Dysfibrinogenemia	ND	↓ Functional fibrinogen; ↑ thrombin time	Heparin, recent birth, liver disease
Excess lipoprotein (a)	10	↑ Lipoprotein (a) level	–
Excess plasminogen activator inhibitor-1 (PAI-1)	ND 10	↑ PAI-1 *PAI* 4G/4G polymorphism (promoter)	Wide range of normal values and diurnal variation
Thrombomodulin deficiency	ND (rare)	↓ Thrombomodulin	–
Plasminogen deficiency	ND	↓ Plasminogen	–
Acquired			
Lupus anticoagulant‡	ND	↑ RVVT (dilute), sensitive PTT, or kaolin clotting time§	Warfarin, heparin, direct oral anticoagulants (DOACs)
Anticardiolipin antibodies‡	ND	IgG or IgM anti-cardiolipin antibodies at moderate–high levels, on ≥2 occasions ≥12 weeks apart	Various infectious diseases
Anti-β_2-glycoprotein I antibodies‡	ND	Anti-β_2-glycoprotein I antibodies present, on ≥2 occasions ≥12 weeks apart	–
Cryoglobulinemia, type I	ND	Type I cryoglobulins present	–
Heparin-induced thrombocytopenia	ND**	• *Screening*: anti-heparin/platelet factor 4 (PF4) antibodies • *Confirmatory*: serotonin release assay	–
Cryofibrinogenemia	ND	Cryofibrinogens present	Acute phase response

*Approximate percentage of general population with the defect.
†Factor V Leiden accounts for ~ 90–95% of patients with APC resistance when the latter is assessed by "second generation" assays, which are not limited by most potential confounding conditions.
‡Antiphospholipid antibodies.
§Confirmed by: (1) failure to correct the prolonged coagulation time in mixing studies; and (2) correction with the addition of excess phospholipids.
**Exposed to heparin or low-molecular-weight heparin.

Table 105.9 Evaluation for thrombophilia. Darkly shaded areas represent first-tier screening tests; lighter shading denotes screening for hyperhomocysteinemia remains controversial and screening for heparin-induced thrombocytopenia is situational. Drugs in **bold** can significantly affect test. The initial laboratory evaluation should also include a complete blood count with differential and platelet count, examination of a peripheral blood smear, ESR, activated partial thromboplastin time (PTT), and hepatic and renal function panels. Testing for antineutrophil cytoplasmic antibodies (ANCA) can be considered for patients with retiform purpura, as ANCA-positive vasculitides occasionally present with minimally inflammatory lesions. Additional inherited causes of thrombophilia include polymorphisms in the genes encoding the endothelial protein C receptor, the protein Z-dependent protease inhibitor, and E-selectin while whole exome sequencing can detect variants in other genes including those that encode serine protease inhibitors (SERPINs). AD, autosomal dominant; CBS, cystathionine β-synthase; MTHFR, methylenetetrahydrofolate reductase; ND, not determined; OCP, oral contraceptive pills; RVVT, Russell viper venom time.

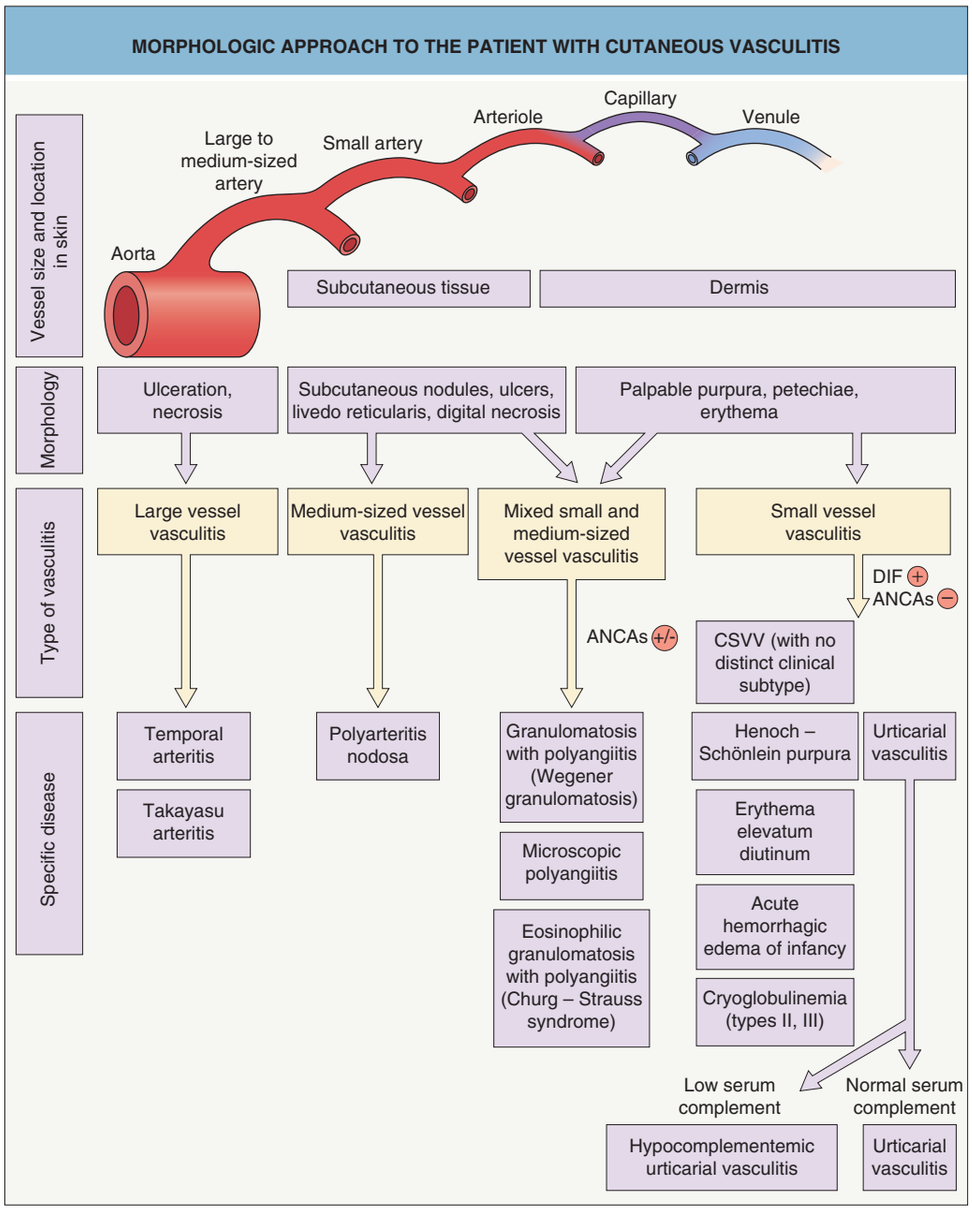

Fig. 24.22 Morphologic approach to the patient with suspected cutaneous vasculitis. ANCAs, antineutrophil cytoplasmic antibodies; CSVV, cutaneous small vessel vasculitis; DIF, direct immunofluorescence.

taken from an ulcer edge or livedo racemosa. Ulcers should be biopsied at their edge and include the peripheral rim of inflammation (if present), as incidental vasculitis can often be found underlying ulcers and is not a diagnostic finding. DIF is best performed on the earliest lesion.

The evaluation of a patient with cutaneous vasculitis is outlined in Fig. 24.16. Additional tests can be performed based upon the suspected associated disease or the suspected vasculitic syndrome (see Fig. 24.16 & Table 24.8). Laboratory tests to determine extent of disease should be repeated with each subsequent episode of vasculitis. Tests for associated diseases are performed at baseline and only repeated if dictated by new information.

Treatment

The first step in the treatment of patients with cutaneous vasculitis is to determine whether the disease is either primary or secondary to an underlying condition (e.g. infection, drug exposure) that can be treated or, in the case of medications, discontinued. Treatment is then based on the extent and severity of cutaneous and systemic involvement. Specific and detailed therapeutic approaches are discussed in conjunction with each disease and are outlined in Table 24.10.

For additional online figures and references visit www.expertconsult.com

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Table 23.3: Classification of vasculitis

I. **Large vessel vasculitis** (aorta and its branching arteries):
   a. Giant cell arteritis
   b. Takayasu's arteritis

II. **Medium-sized vessel vasculitis** (medium-sized and small arteries and veins):
   a. Polyarteritis nodosa
   b. Kawasaki's disease
   c. Cutaneous polyarteritis nodosa
   d. Nodular vasculitis

III. **Small vessel vasculitis** (post-capillary venules, arterioles, and rarely capillaries):
   a. Systemic ANCA-associated vasculitis:
      i. Wegener's granulomatosis
      ii. Churg–Strauss disease
      iii. Microscopic polyangiitis
      iv. Drug-induced (e.g. propylthiouracil)
   b. Henoch–Schönlein purpura
   c. Cryoglobulinemic vasculitis
   d. Cutaneous small vessel vasculitis (leukocytoclastic vasculitis):
      i. Idiopathic
      ii. Secondary forms (infection, malignancy, drugs, etc.)
   e. Other syndromes:
      i. Acute hemorrhagic edema of infancy
      ii. Urticarial vasculitis
      iii. Serum sickness
      iv. Erythema elevatum diutinum
      v. Granuloma faciale
      vi. Eosinophilic vasculitis

Table 23.4: Secondary causes of cutaneous vasculitis

1. Drugs:

- Sulfonamides, minocycline, penicillins, cefaclor, thiazides, NSAIDs, barbiturates, diphenylhydantoin, gold salts, allopurinol, phenothiazines, propylthiouracil, methimazole, radiocontrast media, streptokinase, granulocyte colony stimulating factor (G-CSF), infliximab, interferon-alpha, indinavir, efavirenz
- Hepatitis B, influenza, measles, and typhoid vaccines

2. Infections:

- Streptococcal and staphylococcal infections, syphilis, tuberculosis, brucellosis, leprosy, meningococcosis, gonorrhoea
- Hepatitis A, B, and C infection; herpes simplex; cytomegalovirus, Epstein-Barr virus, HIV, parvovirus B19, and varicella infection
- Rickettsioses
- Candidiasis

3. Malignancies:

- *Hematologic malignancies*: Leukemia, Hodgkin's and non-Hodgkin's lymphoma, multiple myeloma, Waldenström's macroglobulinemia, and myelodysplastic syndrome.
- *Solid organ malignancies*: Carcinomas of the prostate, breast, and colon, ovary; squamous cell carcinoma of the lung; renal cancer

4. Autoimmune diseases:

SLE, rheumatoid arthritis, Sjögren's syndrome, dermatomyositis and polymyositis, relapsing polychondritis

5. Other systemic diseases:

Alpha 1-antitrypsin deficiency, Behçet's disease, congenital deficiency of complement components, chronic active hepatitis, cystic fibrosis, inflammatory bowel disease, intestinal bypass surgery, primary biliary cirrhosis, sarcoidosis, Wiskott–Aldrich syndrome

Table 37.1 Classification of mastocytosis according to WHO recommendations

Cutaneous mastocytosis
(a) Solitary mastocytoma
(b) Maculopapular cutaneous mastocytosis/urticaria pigmentosa
(c) Telangiectasia macularis eruptiva perstans
(d) Diffuse cutaneous mastocytosis
Indolent systemic mastocytosis (ISM)
Systemic mastocytosis with associated clonal hematologic disease (SM-AHNMD)
Aggressive systemic mastocytosis (ASM)
Mast cell leukemia (MCL)
Mast cell sarcoma (MCS)
Extracutaneous mastocytoma

(systemic mastocytosis) and exists for the lifetime of the

Table 11.1 World Health Organization classification of mastocytosis[a]

	Skin lesions
Cutaneous mastocytosis (CM)	+
Urticaria pigmentosa (UP)/ maculopapular CM (MPCM)	–
Diffuse CM	–
Mastocytoma of skin	
Indolent systemic mastocytosis (SM)	+
Smoldering SM	+
Isolated bone marrow mastocytosis	–/+
Systemic mastocytosis with associated clonal hematological non-mast cell lineage disease (SM-AHNMD)	–
Aggressive systemic mastocytosis (ASM)	–/+
Lymphadenopathic mastocytosis with eosinophilia	
Mast cell leukemia (MCL)	–
Aleukemic MCL	
Mast cell sarcoma (MCS)	–
Extracutaneous mastocytoma	–

TABLE 26-3 Detailed Classification of Histiocytoses

Langerhans cell disease	
Xanthogranuloma family	Juvenile xanthogranuloma
	Papular xanthoma
	Scalloped cell xanthogranuloma
	Xanthoma disseminatum
	Reticulohistiocytoma
	Multicentric reticulohistiocytosis
	Spindle cell xanthogranuloma
	Progressive nodular histiocytosis
	Benign cephalic histiocytosis
	Generalized eruptive histiocytosis
Other histiocytic disorders	Indeterminate cell histiocytosis
	Dermal dendrocytoma
	Rosai-Dorfman disease
	Histiocytic sarcoma
Xanthomas associated with lipid abnormalities	Eruptive
	Tuberous
	Tendon
	Plane
	Xanthelasma
Normolipemic xanthomas	Necrobiotic xanthogranuloma
	Diffuse normolipemic plane xanthoma
	Verruciform xanthoma
	Miscellaneous

TABLE 1 Historical classification of histiocytic disorders

Dendritic cell related
Langerhans cell histiocytosis
Juvenile xanthogranuloma/Erdheim–Chester disease
Macrophage related
Hemophagocytic syndromes
Primary hemophagocytic lymphohistiocytosis
Secondary hemophagocytic syndromes
Rosai–Dorfman disease
Malignant diseases
Monocyte-related leukemias
Extramedullary monocytic tumor (myeloid sarcoma)
Macrophage-related histiocytic sarcoma
Dendritic cell malignancy (malignant histiocytosis)

Note: Adapted from Favara et al.[1]

disease (Letterer–Siwe disease), or isolated or multifocal bone lesions (eosinophilic granuloma). Pathologists in the 1950s noted histologic similarity of biopsies from patients with these conditions and proposed a unifying hypothesis that these clinically distinct syndromes represent a common pathological entity, "Histiocytosis X". Subsequently, Nezelof and colleagues identified Birbeck granules' pathologic histiocytes of LCH lesions (Figure 1), a feature at that time was thought to be shared only with epidermal Langerhans cells.

LCH lesions are granulomatous lesions consisting of pathologic "Langerhans cells" (LCs), lymphocytes (primarily T-cells), eosinophils,

TABLE 2 Proposed revised classification of histiocytocytic disorders

L group	LCH
	Intermediate-cell histiocytosis (ICH)
	Erdheim–Chester disease (ECD)
	Mixed LCH/ECD
C group	Cutaneous non-LCH
	-Xanthomatous granuloma (XG) family: includes JXG
	-Non-XG family includes cutaneous RDD
	Cutaneous non-LCH with major systemic component
	-XG family: xanthoma disseminatum
	-Non-XG: multicentric reticulohistiocytosis
R group	Familial RDD
	Sporadic RDD
	-Classic RDD
	-Extranodal RDD
	-RDD with neoplasia or immune disease
	-Unclassified
M group	Primary malignant histiocytosis
	Secondary malignant histiocytosis
H group	Primary HLH
	Secondary HLH (non-Mendelian)
	HLH of unknown/uncertain origin

Note: Adapted from Emile et al.[2]

Abbreviations: ECD, Erdheim–Chester disease; HLH, hemophagocytic lymphohistiocytosis; JXG, juvenile xanthogranuloma; LCH, Langerhans cell histiocytosis; RDD, Rosai–Dorfman disease.

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CLASSIFICATION OF B-CELL LYMPHOMAS WITH PRIMARY CUTANEOUS MANIFESTATIONS	
WHO-EORTC 2005	**WHO 2008/2016**
• Primary cutaneous follicle center lymphoma	• Primary cutaneous follicle center lymphoma
• Primary cutaneous marginal zone B-cell lymphoma*	• Extranodal marginal zone lymphoma of *mucosa-associated lymphoid tissue* – MALT lymphoma
• Primary cutaneous diffuse large B-cell lymphoma, leg type	• Primary cutaneous diffuse large B-cell lymphoma, leg type
• Primary cutaneous diffuse large B-cell lymphoma, other	• Diffuse large B-cell lymphoma, NOS
• Intravascular diffuse large B-cell lymphoma	• Intravascular diffuse large B-cell lymphoma
*Includes cases previously designated as primary cutaneous immunocytoma and primary cutaneous plasmacytoma.	

Table 119.1 Classification of B-cell lymphomas with primary cutaneous manifestations – WHO-EORTC 2005 and WHO 2008/2016. NOS, not otherwise specified.

Table 1 World Health Organization/European Organization for Research and Treatment of Cancer (WHO/EORTC) classification of cutaneous lymphoma and the WHO classification (2018)

WHO-EORTC classification (2018)	WHO classification (2018)
Cutaneous T-cell and NK-cell lymphomas	**Mature T-cell and NK-cell lymphomas**
Mycosis fungoides	Mycosis fungoides
Mycosis fungoides variants and subtypes Folliculotropic Pagetoid reticulosis Granulomatous slack skin	Mycosis fungoides variants and subtypes Folliculotropic Pagetoid reticulosis Granulomatous slack skin
Sézary syndrome	Sézary syndrome
Adult T-cell leukemia/lymphoma (HTLV1$^+$)	Adult T-cell leukemia/lymphoma (HTLV1$^+$)
Primary cutaneous CD30$^+$ lymphoproliferative disorders: Lymphomatoid papulosis Primary cutaneous anaplastic large-cell lymphoma	Primary cutaneous CD30$^+$ lymphoproliferative disorders: Lymphomatoid papulosis Primary cutaneous anaplastic large-cell lymphoma
Subcutaneous panniculitis-like T-cell lymphoma	Subcutaneous panniculitis-like T-cell lymphoma
Extranodal NK/T cell lymphoma, nasal type	Extranodal NK-/T-cell lymphoma, nasal type
Primary cutaneous peripheral T-cell lymphoma Primary cutaneous aggressive epidermotropic CD8$^+$ cytotoxic T-cell lymphoma (provisional) Primary cutaneous CD4$^+$ small to medium-sized T-cell lymphoproliferative disorder (provisional) Primary cutaneous acral CD8$^+$ T-cell lymphoma (provisional) Cutaneous γ/δ T-cell lymphoma	Primary cutaneous peripheral T-cell lymphoma Primary cutaneous aggressive epidermotropic CD8$^+$ cytotoxic T-cell lymphoma (provisional) Primary cutaneous CD4$^+$ small to medium-sized T-cell lymphoproliferative disorder (provisional) Primary cutaneous acral CD8$^+$ T-cell lymphoma (provisional) Cutaneous γ/δ T-cell lymphoma
Cutaneous B-cell lymphomas	**Mature B-cell neoplasias**
Primary cutaneous follicle center lymphoma	Primary cutaneous follicle center lymphoma
Primary cutaneous marginal zone B-cell lymphoma	Primary cutaneous marginal zone B-cell lymphoma (mucosa-associated lymphoid tissue)
Primary cutaneous diffuse large B-cell lymphoma, leg type	Primary cutaneous diffuse large B-cell lymphoma, leg type Primary cutaneous diffuse large B-cell lymphoma, not otherwise specified
Intravascular large cell lymphoma	Intravascular large cell lymphoma
Epstein–Barr virus-positive mucocutaneous ulcer	Epstein–Barr virus-positive mucocutaneous ulcer
Precursor neoplasms	**Precursor neoplasms**
Blastic plasmacytoid dendritic cell neoplasm	Blastic plasmacytoid dendritic cell neoplasm

lymphomas that make the diagnosis of cutaneous lymphomas challenging. Therefore, the diagnosis is always based on a synthesis of clinical, histo- and cytomorphological, immunophenotypic, and molecular characteristics, which allow adequate classification (Kempf and Mitteldorf 2015; Stadler et al. 2013). The staging to assess the clinical spread of the disease is largely based on the tumor, node, metastasis (TNM) system used in oncology, which in its original form was, however, only conditionally suitable for cutaneous lymphomas and was therefore adapted for cutaneous lymphomas (Tables 2, 3, 4, and 5) (Kim et al. 2007; Olsen et al. 2007). Diagnostic workup is ideally performed at specialized centers and is usually interdisciplinary with oncologists, hematologists, radiologists, and dermatopathologists/hematopathologists.

The following examination is recommended if a cutaneous lymphoma is suspected:

History and Clinical Findings Skin symptoms, lymph node involvement, enlargement of tonsils, liver, and spleen.

Skin Biopsies of Adequate Size These should be partly fixed in formalin and partly frozen in liquid nitrogen. In the case of pathologically enlarged lymph nodes, lymph node biopsy should also be performed; especially in the case of high-grade malignant T- and B-cell lymphomas (exception: lymphomatoid papulosis, marginal zone lymphoma), and NK-cell lymphomas, bone marrow biopsy should be carried out.

Laboratory Tests Blood status, liver and kidney function, and lactate dehydrogenase are important. In the case of suspected leukemic variants, fluorescence-activated cell sorting (FACS) analysis (T- and B-cell subset), serum electrophoresis for B-cell lymphomas, and immune electrophoresis are recommended.

Radiological Examinations X-ray imaging of the thorax, ultrasound, and computed tomography of the liver, spleen, and lymph node stations not accessible by palpation should be carried out. Nowadays, PET-CT is often performed.

INTRODUCTION

The diagnosis and classification of cutaneous lymphomas remain one of the most challenging areas of dermatopathology. At the time the last edition of this book was published, the WHO classification of tumors of hematopoietic and lymphoid tissues based on the REAL classification of lymphomas was being published.[1,2] This classification included some cutaneous lymphomas as subgroups within the main classification framework. At the same time, the Project Group of the European Organization for Research and Treatment of Cancer published a classification of cutaneous lymphomas which came to be known as the EORTC classification of cutaneous lymphomas.[3] Those two classifications were based on a combination of histological, clinical, immunohistological, and cytogenetic features. There were certain differences in terminology and emphasis between these two classifications and the appropriateness of one or other of the classifications was debated in the literature.[4–12] The EORTC classification included only those lymphomas which appeared to be unique to the skin. The REAL/WHO classification had the advantage of including other lymphomas which involved the skin secondarily or rarely as 'primary' cutaneous lymphomas.[13] Other proposals were made for a stand-alone classification of cutaneous lymphomas based on the REAL/WHO classification.[13,14]

Recently, a consensus classification of cutaneous lymphomas, the WHO/EORTC classification of cutaneous lymphomas, has been published[15,16] and critiqued in the literature.[16–18] This compromise classification defines 'primary cutaneous lymphomas' as lymphomas that present in skin with no evidence of extracutaneous disease at the time of diagnosis. There are some inherent problems with this strict definition of primary cutaneous lymphoma as some putative primary cutaneous lymphomas are associated with systemic spread to other systems at the time of presentation, e.g. Sézary syndrome and adult T-cell leukemia/lymphoma. The definition does not encompass secondary cutaneous lymphomas. Nevertheless this classification is widely used and will be used as the framework for the discussion in this chapter (see **Table 41.1**). Other lymphoma entities which only rarely present in the skin or do so as part of systemic spread of lymphoma from another site are classified according to the general WHO classification of lymphomas.

The current diagnosis of lymphomas requires some knowledge of lymphocyte ontogeny and techniques which are used to demonstrate such aspects as cell phenotype, clonality of a proliferation, and cytogenetic features including the presence of viral genetic material. Most laboratories are now able to perform these techniques, including immunohistochemistry, in-situ hybridization, polymerase chain reaction technology (PCR), and fluorescence in-situ hybridization (FISH).[19–22] Flow cytometry is also used for immunophenotyping[23] but

Table 41.1 The Classification of cutaneous lymphomas used in this chapter (based on the WHO–EORTC classification)

Cutaneous T-cell and NK-cell lymphomas
Mycosis fungoides and subtypes
Folliculotropic mycosis fungoides
Pagetoid reticulosis
Granulomatous slack skin
Hydroa vacciniforme-like lymphoma
Sézary syndrome
Adult T-cell leukemia/lymphoma
Primary cutaneous $CD30^+$ lymphoproliferative disorders
Primary cutaneous anaplastic large cell lymphoma
Lymphomatoid papulosis
Subcutaneous panniculitis-like T-cell lymphoma
Extranodal NK/T-cell lymphoma, nasal type
Primary cutaneous peripheral T-cell lymphoma, unspecified
Primary cutaneous aggressive epidermotropic $CD8^+$ T-cell lymphoma (provisional)
Cutaneous γ/δ T-cell lymphoma (provisional)
Primary cutaneous $CD4^+$ small/medium pleomorphic T-cell lymphoma (provisional)
Cutaneous B-cell lymphomas
Primary cutaneous marginal zone B-cell lymphoma
Primary cutaneous follicle center cell lymphoma
Primary cutaneous diffuse large B-cell lymphoma, leg type
Primary cutaneous diffuse large B-cell lymphoma, other
Intravascular large B-cell lymphoma
Plasmablastic lymphoma
T-cell/histiocyte-rich B-cell lymphoma
Lymphomatoid granulomatosis
Precursor hematologic neoplasm
Blastic plasmacytoid dendritic cell neoplasm
Other T/NK-cell lymphomas that may involve the skin
Precursor T-lymphoblastic lymphoma/leukemia
T-cell prolymphocytic leukemia
Angioimmunoblastic T-cell lymphoma
Primary systemic anaplastic large cell lymphoma
Intravascular T- and NK-cell lymphoma
Aggressive NK-cell leukemia
Other T/NK-cell lymphomas and leukemias
Other B-cell lymphomas that may involve the skin
Precursor B-lymphoblastic leukemia/lymphoma
Chronic lymphocytic leukemia/small lymphocytic lymphoma
Mantle cell lymphoma
Primary effusion lymphoma
Lymphoplasmacytic lymphoma/Waldenström's macroglobulinemia
Burkitt and Burkitt-like lymphoma
Plasmacytoma and secondary myeloma
Other lymphomas
Hodgkin lymphoma
Cutaneous infiltrates of leukemias
Myeloid leukemias, myeloproliferative diseases and myelodysplastic syndromes
Lymphoid hyperplasia mimicking primary lymphoma
Lymphoid hyperplasia simulating B-cell lymphoma
Lymphomatoid drug reactions
Reactions resembling $CD30^+$ lymphoproliferative disorders
Pseudolymphomatous folliculitis
Jessner's lymphocytic infiltrate
Acral pseudolymphomatous angiokeratoma
Cutaneous $CD8^+$ T-cell infiltrates in HIV/AIDS
Cutaneous infiltrates in post-transplant lymphoproliferative disorders
Other lymphoproliferative disorders associated with immunosuppression
Methotrexate-associated lymphoproliferative disorders
Cutaneous infiltrates in HIV/AIDS
Miscellaneous
Extramedullary hematopoiesis

Table 1.2 Revised 2018 WHO–EORTC classification of primary cutaneous lymphomas

Cutaneous T-cell lymphomas
Mycosis fungoides
Folliculotropic mycosis fungoides
Pagetoid reticulosis
Granulomatous slack skin
Sézary syndrome
Adult T-cell leukemia/lymphoma
Primary cutaneous CD30$^+$ lymphoproliferative disorders
Cutaneous anaplastic large cell lymphoma
Lymphomatoid papulosis
Subcutaneous panniculitis-like T-cell lymphoma
Extranodal NK/T-cell lymphoma, nasal type
Chronic active EBV infection
Primary cutaneous peripheral T-cell lymphoma, rare subtypes
Primary cutaneous γ/δ T-cell lymphoma
CD8$^+$ aggressive epidermotropic cytotoxic T-cell lymphoma
Primary cutaneous CD4$^+$ small/medium T-cell lymphoproliferative disorder
Primary cutaneous acral CD8$^+$ T-cell lymphoma
Primary cutaneous peripheral T-cell lymphoma, not otherwise specified
Cutaneous B-cell lymphomas
Primary cutaneous marginal zone lymphoma
Primary cutaneous follicle center lymphoma
Primary cutaneous diffuse large cell lymphoma, leg type
EBV$^+$ mucocutaneous ulcer
Intravascular large B-cell lymphoma

Adapted from Willemze *et al.* [2] (American Society of Hematology). Provisional entities are in italic.

Table 1. WHO/EORTC classification of cutaneous lymphomas

WHO/EORTC classification of cutaneous lymphomas	WHO classification for lymphoid tissues† (ICD-O Code)
Cutaneous T-cell and NK-cell lymphomas	Mature T-cell and NK-cell neoplasms
Mycosis fungoides (MF)	Mycosis fungoides (MF) (9700/3)
MF variants and subtypes	MF variants and subtypes
Folliculotropic MF	Folliculotropic MF
Pagetoid reticulosis	Pagetoid reticulosis
Granulomatous slack skin	Granulomatous slack skin
Sézary syndrome	Sézary syndrome (9701/3)
Adult T-cell leukaemia/lymphoma	Adult T-cell leukaemia/lymphoma (9827/3)
Primary cutaneous CD30+ lymphoproliferative disorders	Primary cutaneous CD30+ T-cell lymphoproliferative disorders
Primary cutaneous anaplastic large cell lymphoma	Primary cutaneous anaplastic large cell lymphoma (9718/3)
Lymphomatoid papulosis	Lymphomatoid papulosis (9718/1)
Subcutaneous panniculitis-like T-cell lymphoma*	Subcutaneous panniculitis-like T-cell lymphoma* (9708/3)
Extranodal NK/T-cell lymphoma, nasal type	Extranodal NK/T-cell lymphoma, nasal type (9719/3)
Primary cutaneous peripheral T-cell lymphoma, unspecified	Primary cutaneous peripheral T-cell lymphoma, rare subtypes
Primary cutaneous aggressive epidermotropic CD8+ T-cell lymphoma (provisional)	Primary cutaneous CD8+ aggressive epidermotropic cytotoxic T-cell lymphoma (provisional) (9709/3)
Cutaneous γ/δ+ T-cell lymphoma (provisional)	Primary cutaneous γ/δ T-cell lymphoma (9726/3)‡
Primary cutaneous CD4+ small/medium-sized pleomorphic T-cell lymphoma (provisional)	Primary cutaneous CD4+ small/medium T-cell lymphoma (provisional) (9709/3)
Cutaneous B-cell lymphomas	Mature B-cell neoplasms
Primary cutaneous marginal zone B-cell lymphoma	Extranodal marginal zone lymphoma of mucosa-associated lymphoid tissue (MALT lymphoma) (9699/3)
Primary cutaneous immunocytoma	
Primary cutaneous plasmacytoma	
Follicular hyperplasia with monotypic plasma cells	
Primary cutaneous follicle centre lymphoma	Primary cutaneous follicle centre lymphoma (9597/3)
Growth patterns: follicular, follicular and diffuse, diffuse	Diffuse large B-cell lymphoma, NOS (9680/3)
Primary cutaneous diffuse large B-cell lymphoma, leg type	Primary cutaneous diffuse large B-cell lymphoma, leg type (9680/3)
Primary cutaneous diffuse large B-cell lymphoma, other	Intravascular large B-cell lymphoma (9712/3)
Primary cutaneous intravascular large B-cell lymphoma	
Precursor haematological neoplasm	Precursor neoplasms
CD4+/CD56+ haematodermic neoplasm (formerly blastic NK cell lymphoma)	Blastic plasmacytoid dendritic cell neoplasm (9727/3)

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Table 1.1 Revised 4th edition of the WHO *Classification of Tumours of Haematopoietic and Lymphoid Tissues* (2017)

Myeloproliferative neoplasms
Chronic myeloid leukemia, *BCR-ABL1*–positive
Chronic neutrophilic leukemia
Polycythemia vera
Primary myelofibrosis
Essential thrombocythemia
Chronic eosinophilic leukemia, NOS
Myeloproliferative neoplasm, unclassifiable

Mastocytosis
Cutaneous mastocytosis
Systemic mastocytosis
Indolent systemic mastocytosis
Mast cell sarcoma

Myeloid/lymphoid neoplasms with eosinophilia and gene rearrangement
Myeloid/lymphoid neoplasms with *PDGFRA* rearrangement
Myeloid/lymphoid neoplasms with *PDGFRB* rearrangement
Myeloid/lymphoid neoplasms with *FGFR1* rearrangement
Myeloid/lymphoid neoplasms with PCM1-JAK2 9968/3

Myelodysplastic/myeloproliferative neoplasms
Chronic myelomonocytic leukemia
Atypical chronic myeloid leukemia, *BCR-ABL1*–negative
Juvenile myelomonocytic leukemia
Myelodysplastic/myeloproliferative neoplasm with ring sideroblasts and thrombocytosis
Myelodysplastic/myeloproliferative neoplasm, unclassifiable

Myelodysplastic syndromes
Myelodysplastic syndrome with single lineage dysplasia
Myelodysplastic syndrome with ring sideroblasts and single lineage dysplasia
Myelodysplastic syndrome with ring sideroblasts and multilineage dysplasia
Myelodysplastic syndrome with multilineage dysplasia
Myelodysplastic syndrome with excess blasts
Myelodysplastic syndrome with isolated del(5q)
Myelodysplastic syndrome, unclassifiable
Refractory cytopenia of childhood (provisional entity)

Myeloid neoplasms with germline predisposition
Acute myeloid leukemia with germline *CEBPA* mutation
Myeloid neoplasms with germline *DDX41* mutation
Myeloid neoplasms with germline *RUNX1* mutation
Myeloid neoplasms with germline *ANKRD26* mutation
Myeloid neoplasms with germline *ETV6* mutation
Myeloid neoplasms with germline *GATA2* mutation

Acute myeloid leukemia (AML) and related precursor neoplasms

AML with recurrent genetic abnormalities
AML with t(8;21)(q22;q22.1); *RUNX1-RUNX1T1*
AML with inv.(16)(p13.1q22) or t(16;16)(p13.1;q22); *CBFB-MYH11*
Acute promyelocytic leukemia with *PML-RARA*
AML with t(9;11)(p21.3;q23.3); *KMT2A-MLLT3*
AML with t(6;9)(p23;q34.1); *DEK-NUP214*
AML with inv.(3)(q21.3q26.2) or t(3;3)(q21.3;q26.2); *GATA2*, *MECOM*
AML (megakaryoblastic) with t(1;22)(p13.3;q13.1); *RBM15-MKL1*
AML with BCR-ABL1 (provisional entity)
AML with mutated NPM1
AML with biallelic mutation of *CEBPA*
AML with mutated RUNX1 (provisional entity)

AML with myelodysplasia-related changes

Therapy-related myeloid neoplasms

Acute myeloid leukaemia, NOS
AML with minimal differentiation
AML without maturation
AML with maturation
Acute myelomonocytic leukemia
Acute monoblastic and monocytic leukemia
Pure erythroid leukemia
Acute megakaryoblastic leukemia
Acute basophilic leukemia
Acute panmyelosis with myelofibrosis

Myeloid sarcoma (may present primary in the skin)

Myeloid proliferations associated with Down syndrome
Transient abnormal myelopoiesis associated with Down syndrome
Myeloid leukemia associated with Down syndrome

Blastic plasmacytoid dendritic cell neoplasm (often presents primary in the skin)

Acute leukemias of ambiguous lineage
Acute undifferentiated leukemia
Mixed-phenotype acute leukemia with t(9;22)(q34.1;q11.2); *BCR-ABL1*
Mixed-phenotype acute leukemia with t(v;11q23.3); *KMT2A*-rearranged
Mixed-phenotype acute leukemia, B/myeloid, NOS
Mixed-phenotype acute leukemia, T/ myeloid, NOS
Mixed-phenotype acute leukemia, NOS, rare types
Acute leukemias of ambiguous lineage, NOS

Precursor lymphoid neoplasms
B-lymphoblastic leukemia/lymphoma, NOS (may present primary in the skin)
B-lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities
B-lymphoblastic leukemia/lymphoma with t(9;22)(q34.1;q11.2); *BCR-ABL1*
B-lymphoblastic leukemia/lymphoma with t(v;11q23.3); *KMT2A*-rearranged
B-lymphoblastic leukemia/lymphoma with t(12;21)(p13.2;q22.1); *ETV6-RUNX1*
B-lymphoblastic leukemia/lymphoma with hyperdiploidy
B-lymphoblastic leukemia/lymphoma with hypodiploidy (hypodiploid ALL)
B-lymphoblastic leukemia/lymphoma with t(5;14)(q31.1;q32.1); IGH/*IL3*
B-lymphoblastic leukemia/lymphoma with t(1;19)(q23;p13.3); *TCF3-PBX1*
B-lymphoblastic leukemia/lymphoma, BCR-ABL1–like (provisional entity)
B-lymphoblastic leukemia/lymphoma with iAMP21
T-lymphoblastic leukemia/lymphoma
Early T-cell precursor acute lymphoblastic leukemia
NK-lymphoblastic leukemia/lymphoma (provisional entity)

Mature B-cell neoplasms
Chronic lymphocytic leukemia (CLL)/small lymphocytic lymphoma
Monoclonal B-cell lymphocytosis
  CLL type
  Non-CLL type
B-cell prolymphocytic leukemia
Splenic marginal zone lymphoma
Hairy cell leukemia
Splenic B-cell lymphoma/leukemia, unclassifiable
  Splenic diffuse red pulp small B-cell lymphoma (provisional entity)
  Hairy cell leukemia variant (provisional entity)
Lymphoplasmacytic lymphoma
Monoclonal gammopathy of undetermined significance, IgM
Heavy chain diseases
  μ Heavy chain disease
  γ Heavy chain disease
  α Heavy chain disease
Plasma cell neoplasms
  Non-IgM monoclonal gammopathy of undetermined significance
  Plasma cell myeloma
    Plasma cell myeloma variants
Plasmacytoma
  Solitary plasmacytoma of bone
  Extraosseous plasmacytoma
Monoclonal immunoglobulin deposition diseases
  Primary amyloidosis

Table 1.1 (*cont'd*)

- Light chain and heavy chain deposition diseases
- Plasma cell neoplasms with associated paraneoplastic syndrome
  - POEMS syndrome
  - TEMPI syndrome (provisional entity)
- *Extranodal marginal zone lymphoma of mucosa-associated lymphoid tissue (MALT lymphoma) (includes primary cutaneous marginal zone lymphoma)*
- Nodal marginal zone lymphoma
  - Pediatric nodal marginal zone lymphoma
- Follicular lymphoma
  - In situ follicular neoplasia
  - Duodenal-type follicular lymphoma 9690/3
- Pediatric-type follicular lymphoma
- Large B-cell lymphoma with IRF4 rearrangement (provisional entity)
- *Primary cutaneous follicle center lymphoma*
- Mantle cell lymphoma
  - Leukemic non-nodal mantle cell lymphoma
  - In situ mantle cell neoplasia
- Diffuse large B-cell lymphoma (DLBCL), NOS
  - Germinal center B-cell subtype
  - Activated B-cell subtype
- T-cell/histiocyte-rich large B-cell lymphoma
- Primary DLBCL of the CNS
- *Primary cutaneous DLBCL, leg type*
- EBV-positive DLBCL, NOS
- *EBV-positive mucocutaneous ulcer*
- DLBCL associated with chronic inflammation
- Lymphomatoid granulomatosis
  - Grade 1 or 2
  - Grade 3
- Primary mediastinal (thymic) large B-cell lymphoma
- *Intravascular large B-cell lymphoma (often presents primary in the skin)*
- ALK-positive large B-cell lymphoma
- Plasmablastic lymphoma
- Primary effusion lymphoma
- HHV8-associated lymphoproliferative disorders
  - Multicentric Castleman disease
  - HHV8-positive DLBCL, NOS
  - HHV8-positive germinotropic lymphoproliferative disorder
- Burkitt lymphoma
- Burkitt-like lymphoma with 11q aberration (provisional entity)
- High-grade B-cell lymphoma
- High-grade B-cell lymphoma with *MYC* and *BCL2* and/or *BCL6* rearrangement
- High-grade B-cell lymphoma, NOS
- B-cell lymphoma, unclassifiable, with features intermediate between DLBCL and classic Hodgkin lymphoma

Mature T- and NK-cell neoplasms

- T-cell prolymphocytic leukemia
- T-cell large granular lymphocytic leukemia
- Chronic lymphoproliferative disorder of NK cells (provisional entity)
- Aggressive NK-cell leukemia
- EBV-positive T-cell lymphoproliferative diseases of childhood
  - Systemic EBV-positive T-cell lymphoma of childhood
  - Chronic active EBV infection of T- and NK-cell type, systemic form
  - *Hydroa vacciniforme-like lymphoproliferative disorder*
  - *Severe mosquito bite allergy*
- *Adult T-cell leukemia/lymphoma (may present primary in the skin)*
- *Extranodal NK/T-cell lymphoma, nasal type (may present primary in the skin)*
- Intestinal T-cell lymphoma
  - Enteropathy-associated T-cell lymphoma
  - Monomorphic epitheliotropic intestinal T-cell lymphoma
  - Intestinal T-cell lymphoma, NOS
  - Indolent T-cell lymphoproliferative disorder of the gastrointestinal tract (provisional entity)
- Hepatosplenic T-cell lymphoma
- *Subcutaneous panniculitis-like T-cell lymphoma*
- *Mycosis fungoides*
- *Sézary syndrome*
- *Primary cutaneous CD30-positive T-cell lymphoproliferative disorders*
  - *Lymphomatoid papulosis*
  - *Primary cutaneous anaplastic large cell lymphoma*
- *Primary cutaneous peripheral T-cell lymphomas, rare subtypes*
  - *Primary cutaneous gamma delta T-cell lymphoma*
  - *Primary cutaneous CD8-positive aggressive epidermotropic cytotoxic T-cell lymphoma (provisional entity)*
  - *Primary cutaneous acral CD8-positive T-cell lymphoma (provisional entity)*
  - *Primary cutaneous CD4-positive small/medium T-cell lymphoproliferative disorder (provisional entity)*
- *Peripheral T-cell lymphoma, NOS (may present primary in the skin)*
- **Angioimmunoblastic T-cell lymphoma and other nodal lymphomas of T follicular helper cell origin**
  - Angioimmunoblastic T-cell lymphoma
  - Follicular T-cell lymphoma
  - Nodal peripheral T-cell lymphoma with T follicular helper phenotype
- Anaplastic large cell lymphoma, ALK-positive
- Anaplastic large cell lymphoma, ALK-negative
- Breast implant-associated anaplastic large cell lymphoma (provisional entity)

Hodgkin lymphomas

- Nodular lymphocyte-predominant Hodgkin lymphoma
- Classic Hodgkin lymphoma
  - Nodular sclerosis classic Hodgkin lymphoma
  - Lymphocyte-rich classic Hodgkin lymphoma
  - Mixed cellularity classic Hodgkin lymphoma
  - Lymphocyte-depleted classic Hodgkin lymphoma

Immunodeficiency-associated lymphoproliferative disorders (may present primary in the skin)

- Lymphoproliferative diseases associated with primary immune disorders
- Lymphomas associated with HIV infection
- Posttransplant lymphoproliferative disorders (PTLDs)
  - Nondestructive PTLDs
  - Plasmacytic hyperplasia PTLD
  - Infectious mononucleosis PTLD
  - Florid follicular hyperplasia PTLD
  - Polymorphic PTLD
  - Monomorphic PTLDs (B- and T/NK-cell types)
  - Classic Hodgkin lymphoma PTLD
  - Other iatrogenic immunodeficiency-associated lymphoproliferative disorders

Histiocytic and dendritic cell neoplasms (may present primary in the skin)

- Histiocytic sarcoma
- Tumors derived from Langerhans cells
  - Langerhans cell histiocytosis
  - Langerhans cell sarcoma
- Indeterminate dendritic cell tumor
- Interdigitating dendritic cell sarcoma
- Follicular dendritic cell sarcoma
  - Inflammatory pseudotumor-like follicular/fibroblastic dendritic cell sarcoma
- Fibroblastic reticular cell tumor
- Disseminated juvenile xanthogranuloma
- Erdheim–Chester disease

Italic, primary cutaneous lymphomas.
Adapted from Swerdlow *et al*. [1].

Table 1.2 The WHO classification of tumours of haematopoietic and lymphoid tissues.

B-cell neoplasms
Precursor B-cell neoplasms
Precursor B-lymphoblastic leukaemia–lymphoma

Mature B-cell neoplasms
Chronic lymphocytic leukaemia/small lymphocytic lymphoma
B-cell prolymphocytic leukaemia
Lymphoplasmacytic lymphoma
Splenic marginal zone lymphoma
Hairy cell leukaemia
Plasma cell myeloma
Solitary plasmacytoma of bone
Extraosseous plasmacytoma
Extranodal marginal zone B-cell lymphoma of mucosa-associated lymphoid tissue (MALT lymphoma)
Nodal marginal zone B-cell lymphoma
Follicular lymphoma
Mantle cell lymphoma
Diffuse large B-cell lymphoma
Mediastinal (thymic) large B-cell lymphoma
Intravascular large B-cell lymphoma
Primary effusion lymphoma
Burkitt lymphoma/leukaemia

B-cell proliferations of uncertain malignant potential
Lymphomatoid granulomatosis
Post-transplant lymphoproliferative disorder, polymorphic

T- and NK-cell neoplasms
Precursor T-cell neoplasms
Precursor T-lymphoblastic leukaemia/lymphoma
Blastic NK-cell lymphoma (neoplasm of uncertain lineage and stage of differentiation)

Mature T- and NK-cell neoplasms
T-cell prolymphocytic leukaemia
T-cell large granular lymphocytic leukaemia
Aggressive NK-cell leukaemia
Adult T-cell leukaemia/lymphoma
Extranodal NK/T-cell lymphoma, nasal type
Enteropathy-type T-cell lymphoma
Hepatosplenic T-cell lymphoma
Subcutaneous panniculitis-like T-cell lymphoma
Mycosis fungoides
Sézary syndrome
Anaplastic large cell lymphoma, primary cutaneous
Peripheral T-cell lymphoma, unspecified
Angioimmunoblastic T-cell lymphoma
Anaplastic large cell lymphoma

T-cell proliferation of uncertain malignant potential
Lymphomatoid papulosis

Hodgkin lymphoma
Nodular lymphocyte-predominant Hodgkin lymphoma
Nodular sclerosis classical Hodgkin lymphoma
Lymphocyte-rich classical Hodgkin lymphoma
Mixed cellularity classical Hodgkin lymphoma
Lymphocyte-depleted classical Hodgkin lymphoma

may rarely present a monoclonal pattern, and a 'germline' or polyclonal pattern may be observed in clear-cut lymphomas. In addition, lack of sensitivity may give rise to negative results in many cases of early cutaneous T- or B-cell lymphoma.

A common pitfall in molecular analysis of cutaneous lymphoproliferative disorders is the presence of 'pseudomonoclonal' rearrangement of the J_H gene in cutaneous lymphoid infiltrates characterized by the presence of only a few B lymphocytes. In fact, it is not uncommon to observe this phenomenon in lesions of cutaneous T-cell lymphoma with reactive B lymphocytes. A good rule is to perform at least two (better three) separate extractions of DNA from different sections of the biopsy tissue, and to run one independent PCR assay for every extraction. In this way, different 'pseudomonoclonal' bands will be observed, thus showing overall a polyclonal pattern.

Molecular analyses are also widely used in haematology to identify particular genetic aberrations characteristic of a specific clinicopathological entity, such as the interchromosomal 14;18 translocation typical of nodal follicular lymphoma or the t(2;5) of nodal anaplastic large cell lymphoma. Unfortunately, as yet only a few specific genetic alterations have been identified in primary cutaneous lymphomas.

Other methods in the study of cutaneous lymphoid infiltrates

Fluorescence *in situ* hybridization technique

Chromosomal abnormalities are becoming increasingly important in the study and classification of haematopoietic neoplasms. In recent years, new methods have been developed for evaluation of tissue specimens fixed in formalin and embedded in paraffin, allowing the investigation of routine biopsy samples and archive material. The fluorescence *in situ* hybridization (FISH) technique is based on the same principle as the Southern blot method, relying on the annealing of single-stranded DNA to complementary DNA.

Depending on the probes selected, the FISH method can be used to detect different types of chromosomal abnormalities, including monosomy, trisomy and other aneuploidies, as well as translocations and deletions. The use of FISH applied to routinely processed tissue specimens identified a hitherto unknown t(14;18)(q32;q21) involving the J_H and MALT1 genes in a subset of cases of primary cutaneous marginal zone B-cell lymphomas [15].

Microarrays

The recent development of techniques of comparative genomic hybridization, and the identification of over 19 000 human genes by the Human Genome Project, allows one to

Table 1 WHO classification of mature B-cell, T-cell and NK-cell neoplasms (2008)

Precursor lymphoid neoplasms
B lymphoblastic leukaemia/lymphoma NOS
B lymphoblastic leukaemia/lymphoma with recurrent genetic abnormalities
B lymphoblastic leukaemia/lymphoma with t(9;22); bcr-abl1
B lymphoblastic leukaemia/lymphoma with t(v;11q23); MLL rearranged
B lymphoblastic leukaemia/lymphoma with t(12:21); TEL-AML1 & ETV6-RUNX1
B lymphoblastic leukaemia/lymphoma with hyperploidy
B lymphoblastic leukaemia/lymphoma with hypodiploidy
B lymphoblastic leukaemia/lymphoma with t(5;14); IL3-IGH
B lymphoblastic leukaemia/lymphoma with t(1;19); E2A-PBX1 & TCF3-PBX1
T lymphoblastic leukaemia/lymphoma
Mature B-cell neoplasms
Chronic lymphocytic leukaemia/small lymphocytic lymphoma
B-cell prolymphocytic leukaemia
Splenic marginal zone lymphoma
Hairy cell leukaemia
Lymphoplasmacytic lymphoma/Waldenström macroglobulinaemia
Heavy chain disease
Plasma cell myeloma
Solitary plasmacytoma of bone
Extraosseous plasmacytoma
Extranodal marginal zone B-cell lymphoma of mucosa-associated lymphoid tissue (MALT) type
Nodal marginal zone lymphoma
Follicular lymphoma
Primary cutaneous follicular lymphoma
Mantle cell lymphoma
Diffuse large B-cell lymphoma, NOS
(T-cell/histiocyte-rich type; primary CNS type; primary leg skin type and EBV + elderly type)
Diffuse large B-cell lymphoma with chronic inflammation
Lymphomatoid granulomatosis
Primary mediastinal large B-cell lymphoma
Intravascular large B-cell lymphoma
ALK + large B-cell lymphoma
Plasmablastic lymphoma
Large B-cell lymphoma associated with HHV8 + Castleman disease
Primary effusion lymphoma
Burkitt's lymphoma
B-cell lymphoma, unclassifiable, Burkitt's-like
B-cell lymphoma, unclassifiable, Hodgkin's lymphoma-like
Mature T- and NK-cell neoplasms
T-cell prolymphocytic leukaemia
T-cell large granular lymphocytic leukaemia
Chronic lymphoproliferative disorder of NK cells
Aggressive NK cell leukaemia
Systemic EBV + T-cell lymphoproliferative disorder of childhood
Hydroa vacciniforme-like lymphoma
Adult T-cell lymphoma/leukaemia
Extranodal NK/T-cell lymphoma, nasal type
Enteropathy-associated T-cell lymphoma
Hepatosplenic T-cell lymphoma
Subcutaneous panniculitis-like T-cell lymphoma
Mycosis fungoides
Sézary syndrome
Primary cutaneous CD30 + T-cell lymphoproliferative disorder
Primary cutaneous gamma/delta-positive T-cell lymphoma
Peripheral T-cell lymphoma, NOS
Angioimmunoblastic T-cell lymphoma
Anaplastic large cell lymphoma, ALK + type
Anaplastic large cell lymphoma, ALK-type
Hodgkin's lymphoma (Hodgkin's disease)
Nodular lymphocyte-predominant Hodgkin's lymphomas
Classical Hodgkin's lymphoma
Nodular sclerosis Hodgkin's lymphoma
Lymphocyte-rich classical Hodgkin's lymphoma
Mixed cellularity Hodgkin's lymphoma
Lymphocyte depletion Hodgkin's lymphoma
Posttransplant lymphoproliferative disorders (PTLD)
Plasmacytic hyperplasia
Infectious mononucleosis-like PTLD
Polymorphic PTLD
Monomorphic PTLD (B and NK/T-cell types)
Classical HD-type PTLD
Histiocytic and dendritic cell neoplasms
Histiocytic sarcoma
Langerhans cell histiocytosis
Langerhans cell sarcoma
Interdigitating dendritic cell sarcoma
Follicular dendritic cell sarcoma
Fibroblastic reticular cell tumour
Indeterminate dendritic cell sarcoma
Disseminated juvenile xanthogranuloma

small clonal lymphoid populations and the identification of diseases characterised by the involvement of specific anatomical sites or by other clinical features, such as age (Jaffe *et al*, 2008).

Non-Hodgkin's lymphoma (NHL) is the most frequent type of tumour of the head and neck, representing about 75% of lymphomas in this area (Boring *et al*, 1993). Some types of NHL are especially common in this area. The common B-cell neoplasms are precursor B-lymphoblastic leukaemia/lymphoma, chronic lymphocytic leukaemia/small lymphocytic lymphoma, extranodal marginal-zone B-cell lymphoma of MALT, follicular lymphoma, mantle cell lymphoma, diffuse large B-cell lymphoma, and Burkitt's lymphoma/leukaemia. The most frequent T-cell and NK cell NHL is the extranodal NK/T-cell lymphoma, nasal type (Harris *et al*, 1994).

Clinical evaluation

The timing of diagnosis differs considerably depending on the type of lymphoma. HL is diagnosed more frequently in patients 20–30 years of age, whereas NHL is diagnosed more frequently between 70 and 80 years of age (Urquhart and Berg, 2001). In childhood, extranodal NHL may mimic other clinical entities, such as inflammatory disease, polymorphic reticulosis or rhabdomyosarcoma (La Quaglia, 1994). There are no gender differences between HL and NHL in the head and neck. Generalised symptoms, such as fever, weight loss, night sweats, fatigue or pruritus (Urquhart and Berg, 2001), are more frequent in HL than NHL (41% *vs* 27% respectively). Thus, taking into account the minor frequency of HL in the head and neck, systemic symptoms are not common in this area.

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Table 1 Classification of Leukemias

French–American–British (FAB) classification of acute leukemias
M0 Acute myeloid leukemia with minimal evidence of myeloid differentiation
M1 Acute myeloid leukemia without maturation
M2 Acute myeloid leukemia with maturation
M3 Acute promyelocytic leukemia
M4 Acute myelomonocytic leukemia
M5 Acute monocytic leukemia
M6 Acute erythroleukemia
M7 Acute megakaryoblastic leukemia
Acute lymphoblastic leukemias
Chronic leukemias
Chronic myeloid leukemia
Chronic lymphocytic leukemia, B-cell type
Chronic lymphocytic leukemia, T-cell type
T-cell prolymphocytic leukemia
Adult T-cell leukemia/lymphoma
Hairy cell leukemia
Chronic myelomonocytic leukemia

Table 23.1 Classification of precursor lymphoid neoplasms according to the WHO [17]

B-lymphoblastic leukemia/lymphoma, NOS
B-lymphoblastic leukemia/lymphoma with recurrent genetic abnormalities
B-lymphoblastic leukemia/lymphoma with t(9;22)(q34;q11.2); *BCR-ABL1*
B-lymphoblastic leukemia/lymphoma with t(v;11q23); *MLL* rearranged
B-lymphoblastic leukemia/lymphoma with t(12;21)(p13;q22); *TEL-AML1 (ETV6-RUNX1)*
B-lymphoblastic leukemia/lymphoma with hyperdiploidy
B-lymphoblastic leukemia/lymphoma with hypodiploidy (hypodiploid ALL)
B-lymphoblastic leukemia/lymphoma with t(5;14)(q31;q32); *IL3-IGH*
B-lymphoblastic leukemia/lymphoma with t(1;19)(q23;p13.3); *E2A-PBX1 (TCF3-PBX1)*
T-lymphoblastic leukemia/lymphoma

Table 2.4 Historical terms used for mycosis fungoides*

Erythrodermie pityriasique en plaques disseminées
Parakeratosis variegata
Parapsoriasis en plaques
Parapsoriasis lichenoides
Parapsoriasis variegata
Poikiloderma vasculare atrophicans
Premycotic erythema
Prereticulotic poikiloderma
Retiform type of parapsoriasis
Xanthoerythroderma perstans

* Some of these terms are still used today in some centers.

TABLE 2 Classification of cutaneous pseudolymphomas

Nodular pseudolymphomas
Nodular B-cell pseudolymphoma
Borrelia-associated nodular B-cell pseudolymphoma
Nodular T-cell and mixed pseudolymphoma
Nodular CD30+ pseudolymphoma
Pseudolymphomas as simulators of mycosis fungoides or of other CTCLs
Lymphomatoid contact dermatitis
Lymphomatoid drug reaction
Actinic reticuloid
CD8+ T-cell pseudolymphoma in immunodeficiency
Infections as simulators of T-cell lymphomas
Borrelia-associated T-cell pseudolymphoma
Leishmaniasis-associated T-cell pseudolymphoma[a]
Herpesvirus-associated T-cell pseudolymphoma
Syphilis-associated pseudolymphoma
Other Infections als simulators of CTCL
Inflammatory dermatosis as simulators of CTCL
Other pseudolymphomas
T-cell rich angiomatoid pseudolymphoma
Acral pseudolymphomatous angiokeratoma (APA)
T-cell-rich angiomatoid polypoid pseudolymphoma (TRAPP)
Primary cutneous angioplasmocellular hyperplasia
Lymphoplasmacytoid plaque (LPP)
Cutaneous plasmocytosis
Intravascular pseudolymphomas
Benign atypical intravascular (CD30+) lymphoproliferation

[a]Also presents as nodular T-cell pseudolymphoma.

Predominant T-cell pseudolymphomas
Drug induced • Localized (nodular histology, predominantly CD4) • Diffuse (band-like histology)
UV-induced: for example, chronic actinic dermatitis (predominantly CD8, or mixed)
HIV-related (band-like histology, CD8+)
Insect bite reactions
Idiopathic T-cell CPL (nodular or band-like CD4+)
Predominant B cell pseudolymphomas
Borrelia-related cutaneous lymphoid hyperplasia
Varicella-zoster virus-related CPL
Vaccination-induced CLH
CLH secondary to insect bites
Drug-induced (rare)
Idiopathic

HEREDITARY DISORDERS CHARACTERIZED BY CONGENITAL GENERALIZED HYPERTRICHOSIS		
Disorder	**Inheritance (locus; genetic basis)**	**Other key features**
Increased hair is the major feature		
Congenital hypertrichosis lanuginosa	AD	• Fetal vellus hair is not replaced by normal hair and continues to grow • Fine, downy, silvery-gray to blond lanugo hair that may be up to 10 cm in length • Can create a "dog" or "monkey" facial appearance • Involves entire body surface (except for the palms, soles, dorsal surface of distal phalanges, and prepuce) but hair may be shed over the first year of life • Occasional dental anomalies; rarely other associations (e.g. ear defects, glaucoma, pyloric stenosis, photophobia, ID)
Universal hypertrichosis	AD	• Thicker, longer hair most prominent on the back, proximal extremities and the frontal, temporal, and preauricular areas of the face* • Increases during infancy and tends to persist • Considered by some authors to be simply exaggerated normal hairiness that may be familial
Ambras syndrome (hypertrichosis universalis congenita, Ambras type; HTC1)[45–47]	AD (8q22–q24 breakpoints; position effect down-regulates *TRPS1* expression)	• Fine, silky, lightly colored, long hair involving the entire body but primarily the face, ears, and shoulders • Uniformly distributed on the face, including the nose • Persists for life • Minor facial dysmorphism, dental anomalies, supernumerary nipples
Generalized hypertrichosis with extracutaneous features		
X-linked hypertrichosis (congenital generalized hypertrichosis; HTC2)[48]	XLD (Xq27.1; palindrome-mediated interchromosomal insertion; position effect down-regulates *FGF13* expression)	• Curly, shorter, dark hair most prominent on the face and upper body • Anteverted nostrils, prognathism; occasional dental anomalies, deafness • Female carriers may have nevoid hypertrichosis following lines of Blaschko
Congenital generalized hypertrichosis with or without gingival hyperplasia (HCT3)[47,49]	AD or AR (17q24.2–q24.3 microdeletion or microduplication [AD]; position effect down-regulates *SOX9* expression; also *ABCA5* mutations [AR])	• Dark terminal hairs on the peripheral face, central back, and extremities • In addition to gingival hyperplasia, coarse facies, ID, seizures
Cantú syndrome (hypertrichotic osteochondrodysplasia)	AD (12p21.1; *ABCC9* mutation)	• Coarse facies, osteochondrodysplasia, macrosomia at birth, cardiomegaly
Zimmermann–Laband syndrome 1 & 2	1: AD (1q32.2; *KCNH1* mutation) 2: AD (8p21.3; *ATP6V1B2* mutation)	• Gingival hyperplasia, coarse facies, hypoplastic nails and distal phalanges, joint hyperextensibility, splenomegaly, ID
Coffin–Siris syndrome 1–5**	1: AD (6q25.3; *ARID1B* mutation) 2: AD (1p36.11; *ARID1A* mutation) 3: AD (22q11.23; *SMARCB1* mutation) 4: AD (19p13.2; *SMARCA4* mutation) 5: AD (17q21.2; *SMARCE1* mutation)	• Sparse scalp hair, coarse facies, hypoplastic fifth fingernails and toenails, GD, ID
Schinzel–Giedion midface retraction syndrome	AD (18q12.3; *SETBP1* mutation)	• Midfacial vascular stain, midfacial retraction, hyperconvex nails, hypoplastic distal phalanges and dermatoglyphs, GU anomalies, GD, ID
Gorlin–Chaudry–Moss syndrome	AR vs XLD	• Craniofacial dysostosis with midfacial hypoplasia, hypoplastic distal phalanges, ocular and dental anomalies, genital hypoplasia, GD
Adducted thumbs syndrome	AR	• Arthrogryposis, craniosynostosis, myopathy
Barber–Say syndrome	AD (2q37.3; *TWIST2* mutation)	• Lax skin, ectropion, macrostomia, coarse facies, hypoplastic nipples, GD
Amaurosis congenita, cone–rod type, with congenital hypertrichosis‡	AR	• Photophobia, visual impairment due to retinal dystrophy
CAHMR syndrome‡	AR	• *C*ataracts, *h*ypertrichosis, *m*ental *r*etardation

*This pattern of distribution is also observed in prepubertal hypertrichosis (see text).
**Protein products of mutated genes are subunits of the SWI/SNF complex.
‡Single family reported to date.

Table 70.1 Hereditary disorders characterized by congenital generalized hypertrichosis. ABC, ATP-binding cassette transporter; AD, autosomal dominant; AR, autosomal recessive; ARID, AT-rich interaction domain; ATP6V1B2, ATPase H+ transporting V1 subunit B2; FGF, fibroblast growth factor; GD, growth delay; GU, genitourinary; HTC, hypertrichosis; ID, intellectual disability; KCN, potassium voltage-gated channel; SMARC, SWI/SNF related, matrix associated, actin dependent regulator of chromatin; TRPS1, transcriptional repressor GATA binding 1/tricho-rhino-phalangeal syndrome 1 protein; TWIST, twist family BHLH transcription factor 2; XLD, X-linked dominant.

HEREDITARY DISEASES AND CONGENITAL SYNDROMES ASSOCIATED WITH REGIONAL HYPERTRICHOSIS		
Disorder	**Sites of hypertrichosis**	**Other key features**
Dysmorphic syndromes		
Cornelia de Lange syndrome 1–5	• Forehead, lateral face, shoulders, back • Low anterior hairline, synophrys, trichomegaly	• Distinctive facies, upper extremity anomalies, microcephaly, GD, ID, "growling cry" • AD (1,3,4): *NIPBL* > *SMC3*, *RAD21* mutations • XLD (2,5): *SMC1A*, *HDAC8* mutations
Rubinstein–Taybi syndrome 1 & 2	• Lateral face, shoulders, back • Thick eyebrows, trichomegaly	• Midfacial vascular stains, keloids, pilomatricomas • Broad thumbs/halluces, beaked nose, high-arched palate, short stature, ID • AD; *CREBBP* mutation (or deletion) > *EP300* mutation
Disorders with primary cutaneous features		
Porphyrias	• Sun-exposed areas • Favors lateral face in PCT, HEP, VP • Face, trunk, extremities in CEP	• See Ch. 49
Lipodystrophy syndromes (e.g. Berardinelli–Seip syndrome, leprechaunism)	• Face, neck, extremities • Low hairline	• See Ch. 101
Erythrokeratodermia variabilis	• Trunk and extremities	• See Ch. 57
Dystrophic epidermolysis bullosa*	• Areas of previous blistering	• See Ch. 32
Ichthyosis bullosa of Siemens*	• Extremities	• See Ch. 57
Metabolic disorders		
Mucopolysaccharidoses†	• Trunk, extremities	• See Ch. 48
Congenital hypothyroidism	• Back, extremities	• See Ch. 46
Mitochondrial disorders		
Leigh syndrome due to *SURF1* mutations	• Forehead, extremities	• Progressive neurodegeneration
MELAS syndrome	• Lower extremities	• *M*itochondrial *e*ncephalopathy, *l*actic *a*cidosis, and stroke-like episodes
Intrauterine exposures‡		
Fetal hydantoin syndrome	• Face, back, extremities	• Gingival hyperplasia, coarse facies, hypoplastic nails and distal phalanges, GD
Fetal alcohol syndrome	• Face, back, extremities	• Midfacial hypoplasia, GD, CNS anomalies
Morpheaform or sclerodermoid disorders		
MONA (multicentric osteolysis, nodulosis, and arthropathy; previously known as Winchester syndrome)	• Hypertrichosis and hyperpigmentation overlying areas of skin thickening	• Gingival hyperplasia, carpal/tarsal osteolysis, osteoporosis, coarse facies, GD • AR; matrix metalloproteinase-2 gene mutations
H syndrome§,50–52	• Lower trunk and lower extremities (in association with areas of *h*yperpigmentation & induration) • Histologic features in areas of induration include a polyclonal perivascular lymphohistiocytic infiltrate with numerous plasma cells in the dermis and subcutis	• Sensorineural *h*earing loss, short *h*eight, *h*eart anomalies, *h*epatosplenomegaly, scrotal masses, hypergonadotropic *h*ypogonadism, antibody-negative insulin-dependent diabetes mellitus, facial telangiectasias • AR; *SLC29A3* mutations (encodes nucleoside transporter hENT3 which localizes to lysosomes and mitochondria)
Stiff skin syndrome	• Variable	• See Ch. 43
Linear melorheostosis (isolated melorheostosis)¶	• Overlying affected skin and bone	• Hyperostosis ("candle wax" appearance) with overlying scleroderma • Isolated cases due to *LEMD3* mutations

*Hypertrichosis is an uncommon feature.
†Hypertrichosis may also be observed in other lysosomal storage diseases, e.g. Krabbe disease and GM1-gangliosidosis, as well as sialuria.
‡Also minoxidil and diazoxide.
§Allelic with pigmented hypertrichosis with insulin-dependent diabetes mellitus (PHID) syndrome, sinus histiocytosis with massive lymphadenopathy (SHML), and Faisalabad histiocytosis.
¶Hypertrichosis has also been described overlying linear scleroderma in the absence of melorheostosis.

Table 70.2 Hereditary diseases and congenital syndromes associated with regional hypertrichosis. Varying degrees of hypertrichosis (predominantly facial or generalized) have also been observed in children with chromosomal anomalies, e.g. partial trisomy of 1q, 3q, 4p or 17q. *NIPBL*, *SMC1A*, *SMC3*, and *RAD21* encode components of the cohesin complex. *RAD21* and *EP300* encode a histone acetyltransferase and a histone deacetylase, respectively. AD, autosomal dominant; AR, autosomal recessive; CEP, congenital erythropoietic porphyria; GD, growth delay; HEP, hepatoerythropoietic porphyria; ID, intellectual disability; PCT, porphyria cutanea tarda; VP, variegate porphyria; XLD, X-linked dominant.

TABLE 1 | Isolated non-syndromic hereditary hair loss disorders, classified by OMIM naming system.

OMIM Name	Disease	Inheritance	Gene and locus	Phenotype
Hypotrichosis 1 (HYPT1)	Hereditary hypotrichosis simplex type 1 (HHS1)	AD	APCDD1 18p11.22	Normal hair at birth, generalised hypotrichosis progressing from early childhood, with sparse, thin and short hair on scalp and body. Sparing of eyebrow, eyelash and beard hair [4].
Hypotrichosis 2 (HYPT2)	Hereditary hypotrichosis simplex type 2 (HHS2)	AD	CDSN 6p21.3	Normal hair at birth, progressive scalp hair loss from the first decade to diffuse loss by the third decade. Sparse, fine, short scalp hairs. Sparing of eyebrow, eyelash, beard and axillary hair [5].
Hypotrichosis 3 (HYPT3)	Dominant hereditary hypotrichosis 3	AD	KRT74 12q13	Sparse scalp hair from birth, sometimes wiry irregular hair in childhood, with progressive loss to puberty. Sparing of eyebrow, eyelash and facial hair. Normal teeth and nails [6].
Hypotrichosis 4 (HYPT4)	Marie Unna hereditary hypotrichosis 1 (MUHH1)	AD	U2HR 8p21.2	Sparse or absent hair at birth. Slow-growing, variable, coarse, wiry, fragile hair from childhood. Affecting scalp, eyebrows and body hair. Electron microscopy displaying an irregular twisting hair dystrophy and longitudinal grooving [7].
Hypotrichosis 5 (HYPT5)	Marie Unna hereditary hypotrichosis 2 (MUHH2)	AD	EPS8L3 1p13.3	Sparse to absent scalp hair at birth. Wiry, irregular hair in childhood. Thin eyebrows and eyelashes with absence of axillary and pubic hair [8].
Hypotrichosis 6 (HYPT6)	Localised autosomal recessive hypotrichosis 1 (LAH1)	AR	DSG4 18q12	Sparse fragile scalp hairs, affected or absent eyebrows and eyelashes and sparing facial, axillary and pubic hairs. Occasionally hyperkeratotic follicular papules on scalp or other affected areas—an association among LAH phenotypes [9].
Hypotrichosis 7 (HYPT7)	Localised autosomal recessive hypotrichosis 2 (LAH2)	AR	LIPH 3q27.2	Sparse, wiry and twisted to woolly scalp hair. Wide spectrum of body hair phenotype, with individuals affected to different degrees in adulthood [10].
Hypotrichosis 8 (HYPT8)	Localised autosomal recessive hypotrichosis 3 (LAH3)	AR	LPAR6 13q14	Tightly curled, woolly, fragile and slow-growing scalp and trunk hair, and normal to sparse eyebrows and eyelashes [9, 11]. Sparing of beard, and axillary and pubic hairs [11].
Hypotrichosis 9 (HYPT9)	Localised autosomal recessive hypotrichosis 4 (LAH4)	AR	Unknown 10q11.23-q22.3	Thin, sparse hair present from birth on scalp, arms and legs. Sparing of eyebrows and eyelashes, with no other dysmorphic features, no hearing or neurological abnormalities [12].
Hypotrichosis 10 (HYPT10)	Localised autosomal recessive hypotrichosis 5 (LAH5)	AR	Unknown 7p22.3-p21.3	Complete absence of scalp hair from birth. Sparse eyebrow, eyelash, beard and body hair. Papules present on scalp at the termination of hair follicles [13].

(Continues)

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TABLE 1 | (Continued)

OMIM Name	Disease	Inheritance	Gene and locus	Phenotype
Hypotrichosis 11 (HYPT11)	Hereditary hypotrichosis simplex type 3 (HSS3)	AD	SNRPE 1q32.1	Sparse to complete absence of scalp, eyelash, eyebrow and axillary and body hair from birth. Pubic hairs spared [14, 15].
Hypotrichosis 12 (HYPT12)	Hereditary hypotrichosis simplex type 4 (HSS4)	AD	RPL21 13q12.2	Normal hair at birth and progressive hair loss 2–6 months of age, progressing to sparse or complete loss of scalp hair. Generalised thin, fragile, dry and sparse hair; some with affected eyebrows, eyelashes and body hair. Sparing of beard hair [16, 17].
Hypotrichosis 13 (HYPT13)	Hypotrichosis with woolly hair	AD	KRT71 12q13.13	Tightly curled scalp woolly hair from birth with limited length. No hair shaft or follicular abnormalities. Affecting scalp, eyebrow and eyelash hairs [18].
Hypotrichosis 14 (HYPT14)	Hypotrichosis 14	AR	LSS 21q22.3	Sparse to absent lanugo-like scalp hair, short brittle eyelashes and eyebrows, and sparse body hair [19].
HYPTSV	Hypotrichosis and recurrent skin vesicles	AR	DSC3 18q12.1	Sparse to absent scalp hair; absence of eyebrows, eyelashes, axillary and body hair and cutaneous vesicles of < 1 cm in diameter on scalp and skin [20, 21].
—	Digenic autosomal recessive hypotrichosis	AR	CDH3 16q22.1 & Unknown 12q21.2-q22	Sparse to absent scalp hair from birth, with sparse to normal eyebrows and eyelashes and facial, axillary and body hair [22].

Note: Colours denote broad groups of disease phenotypes.
Abbreviations: AD = autosomal dominant, AR = autosomal recessive [23, 24].

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Table 1: Summarizes the types of hereditary sensory autonomic neuropathy (HSAN)[4-6]

Type	Gene	Inheritance	Locus	Clinical features	Age of onset
HSAN IA (most common)	SPTLC1	AD	9q22.2	Predominant loss of pain and temperature Preservation of vibration sense Lancinating pain Variable distal motor involvement	Adult
HSAN IB	Unknown	AD	3p24-p22	Predominant sensory neuropathy (cough and gastroesophageal reflux) rarely foot ulceration	Adult
HSAN IC	RAB7	AD	3q21	Prominent distal motor loss Sensory loss of all qualities Acro-mutilations	Adult
HSAN II	WNK1/HSN2	AR	12p13.3	Predominant sensory loss Mutilation of hands and feet. Acropathy	childhood
HSAN III	IKBKAP Riley Day dysautonomia	AR	9q31	Prominent autonomic dysfunction Absent fungiform papillae Alacrimia Absent sweating	Congenital
HASN IV	NTRK 1 Congenital Insenstivity to pain	1q21	AR	Reduced pain sensation Anhidrosis Episodic fever Mild Mental retardation Cornea lesions and joint deformities	Congenital
HSAN V	NGFB	1p13.1	AR	Painless fracture Joint deformities Insensitivity to pain Normal intelligence	Congenital
HSAN with spastic paraplegia	CCT5	5p15	AR	Prominent sensory neuropathy Mutilating acropathy Spastic paraplegia	Early childhood

TABLE 1. *Hereditary Sensory and Autonomic Neuropathies (HSAN)*

	HSAN I	HSAN II	HSAN III	HSAN IV	HSAN V
Inheritance	AD	AR	AR	AR	?
Age of onset	Second decade or later	Infancy	Birth	Birth	Birth
Insensitivity to pain	++	++	+	++	++
Hidrosis	None	None		Decrease	None
Skin blotching	Absent		+	+	
Fungiform papillae of the tongue	Present	Present	Absent	Present	
Esophageal gastrointestinal involvement	Absent	Sometimes	++	Absent	
Mental retardation	Absent	–/+	+	++	–/+
Muscle hypotonia	Absent	Absent	+	Absent	Absent
Deep tendon reflexes	Absent	Absent	Often absent	None	
Motor nerve conduction velocity	None	None	None	Normal/ decreased	None
Sensory nerve conduction velocity	Low/absent	Low/absent	Low/absent	Low/absent	None
Biopsy finding	Loss of UF > MF	Absent MF	Reduced UF	Absent UF	Absent small MF

AD = autosomal dominant, AR = autosomal recessive, UF = unmyelinated fibers, MF = myelinated fibers.

DISORDERS CHARACTERIZED BY DIFFUSE PIGMENTARY DILUTION IN WHICH THE GENETIC DEFECT IS KNOWN			
Disorder	**Gene**	**Protein product**	**Comments**
Oculocutaneous albinism (OCA) **[~40% of patients have OCA1 and ~50% have OCA2]**			
OCA1A	*TYR*	Tyrosinase	• Complete absence of tyrosinase activity and melanin production • Retention of misfolded tyrosinase protein within the ER
OCA1B	*TYR*	Tyrosinase	• Decreased tyrosinase activity; can produce pheomelanin • Variant with temperature-sensitive tyrosinase (normal activity at 35°C, but diminished at 37°C) • Additional variants: minimal pigment, platinum, yellow
OCA2	*OCA2*	P protein (*OCA2* was previously known as *P*)	• Melanosomal transmembrane protein that is also present in the ER • Possible functions include regulating organelle pH, facilitating vacuolar accumulation of glutathione, and processing/trafficking of tyrosinase
OCA3	*TYRP1*	Tyrosinase-related protein 1*	• TYRP1 stabilizes tyrosinase in mice and humans, and it can function as a DHICA oxidase • Both "mutant" TYRP1 and tyrosinase are retained in the ER and then degraded • Rufous phenotype ≫ brown phenotype; latter seen in OCA2
OCA4	*SLC45A2*	Solute carrier family 45 member 2 (previously known as MATP)	• Variable phenotype, with hair ranging from white to yellow–brown; most common in Asians • Transmembrane transporter with a role in tyrosinase processing and intracellular trafficking of proteins to the melanosome
OCA5	Not known	–	• Locus linked to 4q24
OCA6	*SLC24A5*	Solute carrier family 24 member 5	• Cation exchanger that may be involved in ion transport in melanosomes
OCA7	*C10orf11*	Chromosome 10 open reading frame 11	• Expressed in melanoblasts and melanocytes • Possible role in melanocyte differentiation
Hermansky–Pudlak syndrome (HPS)†			
HPS1	*HPS1*	HPS1 (BLOC-3 subunit 1)	• Defective trafficking of organelle-specific proteins to melanosomes, lysosomes and cytoplasmic granules (including platelet dense granules and lytic granules in cytotoxic T lymphocytes) • Pulmonary fibrosis and granulomatous colitis • Component of BLOC-3 (see Fig. 65.8)
HPS2	*AP3B1*	Adaptor related protein complex 3 β1 subunit	• AP-3 recognizes sorting signals within cytosolic tails of cargo molecules and is involved in the trafficking of proteins from the trans-Golgi network to appropriate organelles (see Fig. 65.8) • Pulmonary fibrosis (some patients) • Abnormal targeting of CD1 may play a role in associated immunodeficiency
HPS3	*HPS3*	HPS3 (BLOC-2 subunit 1)	• Component of BLOC-2
HPS4	*HPS4*	HPS4 (BLOC-3 subunit 2)	• Pulmonary fibrosis • Component of BLOC-3
HPS5	*HPS5*	HPS5 (BLOC-2 subunit 2)	• Component of BLOC-2
HPS6	*HPS6*	HPS6 (BLOC-2 subunit 3)	• Component of BLOC-2
HPS7	*DTNBP1*	Dystrobrevin binding protein 1	• Component of BLOC-1
HPS8	*BLOC1S3*	BLOC1S3 (BLOC-1 subunit 3)	• Component of BLOC-1
HPS9	*BLOC1S6*	BLOC1S6 (BLOC-1 subunit 6)	• Component of BLOC-1
HPS10	*AP3D1*	Adaptor related protein complex 3 δ1 subunit	• See AP-3 above • Neurologic impairment (e.g. seizures), immunodeficiency
Chédiak–Higashi syndrome			
	LYST	Lysosomal trafficking regulator	• Abnormal vesicle trafficking and fission/fusion of lysosome-related organelles result in giant organelles (e.g. melanosomes, neutrophil granules [lysosomes], platelet dense granules)
Griscelli syndrome			
GS1	*MYO5A*	Myosin Va (attaches melanosomes to actin filaments)	• In all three forms, melanosomes are retained in the body of the melanocyte rather than trafficking to the tips of dendrites for transfer to keratinocytes (see Fig. 65.10) • Silvery hair seen in all three forms • Myosin Va expressed in neurons (as well as melanocytes) and dysfunction leads to neurologic abnormalities
GS2	*RAB27A*	RAB27A (melanosomal membrane GTPase that binds melanophilin)	• GTPase also expressed in hematopoietic cells; defective release of granule contents from cytotoxic T cells leads to recurrent infections and hemophagocytic syndrome
GS3	*MLPH*	Melanophilin (links myosin Va and RAB27A)	• Melanophilin only expressed in melanocytes, so only pigmentary dilution in type 3
	MYO5A	Myosin Va F-exon deletion	• F-exon only expressed in melanocytes

*Recognized by Mel-5 antibody.
†Reflecting a founder effect, individuals of Puerto Rican origin have HPS1 and HPS3 (3 : 1 ratio); non-Puerto Ricans most commonly have HPS1, followed by HPS3 and HPS4 with ~70% of patients having one of these three types.

Table 65.1 Disorders characterized by diffuse pigmentary dilution in which the genetic defect is known. See Chapter 66 for details of clinical findings. BLOC-1 promotes endosomal maturation by recruiting the Rab5 GTPase-activating protein Msb3; BLOC-2 targets recycling endosomal tubules to melanosomes for cargo delivery; BLOC-3 functions as a Rab32/38 guanine nucleotide exchange factor that is capable of activating small GTPases (activated Rab 32/38 is needed for transport of tyrosinase and TYRP1 to melanosomes). AP-3, adaptor protein complex 3; BLOC, *b*iogenesis of *l*ysosome-related *o*rganelles *c*omplex; DHICA, 5,6-dihydroxyindole-2-carboxylic acid; ER, endoplasmic reticulum; MATP, membrane associated transporter protein; TYRP, tyrosinase-related protein.

and myopia.

Table 49.3: Xeroderma pigmentosum complementation groups. Adapted from Giordano et al. 2016, (Tables 49.2 and 49.3).[183]

Complementation group	*Protein*	*Photosensitivity*	*Cancer predisposition*	*Neurologic abnormalities*
A	XPA	+++	+++	+++
B	XPB and ERCC3	+++	+++	+++
C	XPC	+ to +++	+++	Rare
D	XPD and ERCC2	+ to +++	– to +++	>50%
E	XPE and DDB2	+ to +++	+	Rare
F	XPF and ERCC4	+ to +++	+	+ to +++
G	XPG and ERCC5	+ to +++	+ to +++	– to +++
Variant	Pol η	+++	+++	Rare

(*DDB2*: DNA damage-binding protein 2; *ERCC*: excision repair cross-complementing; *XP*: xeroderma pigmentosum)

CLASSIFICATION SCHEME FOR APLASIA CUTIS CONGENITA (ACC)			
	Inheritance (Gene)	**Features**	**Associated abnormalities**
Group 1: Scalp ACC without multiple anomalies	Sporadic > AD	Usually at the vertex Membranous Hair collar	**No associated abnormalities** **Isolated abnormalities** (most often associated with membranous ACC): cleft lip and/or palate, tracheoesophageal fistula, high myopia and cone-rod dysfunction, patent ductus arteriosus, intestinal lymphangiectasia, omphalocele, polycystic kidneys, double cervix and uterus, intellectual disability, temporal triangular alopecia
	Often AD (*BMS1*)	Usually at the vertex Irregular and scar-like	
Group 2: Scalp ACC associated with Adams–Oliver syndrome (types 1–6)	AD (*ARHGAP31*, *RBPJ*, *NOTCH1, DLL4*) AR (*DOCK6*, *EOGT*)	Midline scalp Often large, irregular lesions Skull defect Dilated scalp veins	**Adams–Oliver syndrome**: terminal transverse limb defects (e.g. reductions, syndactyly, nail dystrophy), cutis marmorata telangiectatica congenita, cardiac malformations, CNS abnormalities
Group 3: Scalp ACC associated with epidermal and organoid nevi (and, occasionally, large congenital melanocytic nevi)	Sporadic due to mosaic mutations (e.g. *HRAS*, *KRAS*)	Scalp, often unilateral Membranous	**Epidermal nevus or nevus sebaceus**, usually in proximity to the ACC **Large congenital melanocytic nevus**: in a few reports* **Ipsilateral ophthalmologic abnormalities**: corneal opacities, scleral dermoids, eyelid colobomas, retinal dystrophy **CNS abnormalities**: intellectual disability, seizures
Group 4: ACC overlying embryologic malformations	Variable	Scalp, often with a hair collar if overlying a neural tube defect Lumbosacral Chest, abdomen	**Cranial malformations**: cephalocele, heterotopic brain or meningeal tissue, congenital midline porencephaly, holoprosencephaly, craniosynostosis, vascular malformation/aplasia/arteriovenous fistula **Spinal malformations**: occult dysraphism, myelomeningocele **Thoracic malformations**: sternal cleft **Abdominal malformations**: omphalocele, gastroschisis **Multiple malformations associated with generalized, lethal ACC ("systemic" ACC)**: corneal opacities, choanal atresia, hypoplastic lungs, absent greater omentum, intestinal malrotation & atresias, hepatosplenomegaly, skull defects, syndactyly, anonychia
Group 5: ACC associated with fetus papyraceus, placental infarcts, or other ischemic events	Sporadic	Scalp, chest, flanks, axillae and/or extremities (see Fig. 64.17) Multiple, symmetric Stellate/angulated configuration	**Associated fetus papyraceus** due to second-trimester death of twin/triplet or fetal reduction of higher-order pregnancies **Placental infarction**, single umbilical artery **Duodenal, ileal, and/or biliary atresias with intestinal infarction** Volkmann ischemic contracture of the forearm Intracranial hemorrhage/CNS abnormalities Amniotic bands, clubbed hands/feet, nail dystrophy
Group 6: ACC associated with EB	AD or AR, depending on EB type	Lower extremities ("Bart syndrome") or large areas of the trunk and extremities	**Simplex, junctional and dystrophic (including bullous dermolysis of the newborn) types of EB**: blistering of skin and/or mucous membranes, skin fragility, nail dystrophy **In more severe cases (most associated with junctional EB)**: pyloric atresia, ureteral stenosis, renal abnormalities, arthrogryposis, abnormal ears and nose, amniotic bands
Group 7: ACC localized to extremities without blistering	AD or AR	Pretibial areas, extensor forearms, dorsal hands and feet	**Radial dysplasia** (associated with ACC on the extensor forearm)
Group 8: ACC caused by specific teratogens, maternal conditions or intrauterine infections	Not inherited	Scalp with drugs Any site with congenital infections	**Methimazole (thiamazole), carbimazole**: imperforate anus, urachal malformations **Misoprostol** **Low-molecular-weight heparin** **Valproic acid** **Maternal antiphospholipid syndrome** **Intrauterine varicella, herpes simplex or rubella infection**: signs of these infections
Group 9: ACC associated with malformation syndromes	Variable	Variable	See Table 64.4 **Amniotic band sequence and disorganization syndrome** (see text)

*Referred to as "SCALP" syndrome (*s*ebaceous nevus, *C*NS malformations, *A*CC, *l*imbal dermoid, *p*igmented nevus).

Table 64.3 Classification scheme for aplasia cutis congenita (ACC). Bold text in column 4 signifies main topics, followed by details. AD, autosomal dominant; AR, autosomal recessive; *ARHGAP31*, Rho GTPase activating protein 31; *BMS1*, ribosome biogenesis factor; CNS, central nervous system; *DLL4*, Delta-like 4; DOCK6, dedicator of cytokinesis 6; EB, epidermolysis bullosa; *EOGT*, EGF domain-specific O-linked N-acetylglucosamine transferase; *RBPJ*, recombination signal binding protein for immunoglobulin κ J region (transcriptional regulator for the Notch pathway). *Adapted from Frieden IJ. J Am Acad Dermatol. 1986;14:646–60, with permission.*

TABLE 12.1. Nomenclature of the parapsoriases.

Disease entity	Synonyms
Large-plaque parapsoriasis[a,b]	Large-plaque (atrophic) parapsoriasis[c]
	LPAP[c]
	Atrophic parapsoriasis[c]
	Poikilodermatous parapsoriasis[c]
	Parapsoriasis en plaques,[c] large-plaque type
	Parapsoriasis en plaques,[d] atrophic type
	Parapsoriasis en grandes plaques simples
	Parapsoriasis en grandes plaques poikilodermiques
	Lichenoid stage of mycosis fungoides
	Poikilodermic mycosis fungoides
	Prereticulotic dermatitis
	Prereticulotic poikiloderma
	Parapsoriasis en plaques[b,d,e]
	Poikiloderma vasculare atrophicans[e,f]
	Parapsoriasis lichenoides[e,g]
Variant: retiform parapsoriasis[h]	Parapsoriasis variegata[c,g]
	Parakeratosis variegata[c]
	Lichen variegatus
	Parapsoriasis lichenoides[e,g]
Small-plaque parapsoriasis[a,b]	Parapsoriasis en plaque,[d] small-plaque type
	Parapsoriasis en plaques,[d] simple discrete type
	Parapsoriasis en plaques,[d] benign type
	Leopard-spot parapsoriasis
	Chronic superficial dermatitis[e]
	Parapsoriasis en plaques[b,d,e]
Variant: digitate dermatosis[i]	Fingerprint parapsoriasis[i]
Variant: xanthoerythroderma perstans[i,j,k]	
Pityriasis lichenoides[l]	Parapsoriasis guttata[c,e,g]
Acute[l]	Pityriasis lichenoides et varioliformis acuta[c]
	PLEVA[c]
	PLVA[c]
	Mucha-Haberman disease[c]
	Parapsoriasis lichenoides et varioliformis acutea
	Parapsoriasis violiformis
	Parapsoriasis acuta[g]
	Acute guttage parapsoriasis
	Acute pityriasis lichenoides
Chronic[l]	Pityriasis lichenoides chronica[c,g]
	Parapsoriasis lichenoides chronica[g]
	Chronic guttate parapsoriasis

Source: Adapted from Lambert, WC, Everett MA: The nosology of parapsoriasis. *J Am Acad Dermatol* 1981; 5:373–395.

[a] Some authorities consider large-plaque and small-plaque parapsoriasis as a single entity, usually termed *parapsoriasis en plaques*.

[b] The term *Brocq's disease* is sometimes utilized to denote small- and large-plaque parapsoriasis together as a single entity; less commonly it is used to denote one or more of the other parapsoriases.

[c] Preferred synonyms, based on frequent usage.

[d] "Parapsoriasis en plaques" is pronounced with the final "s" silent, misleading some English-speaking authors to misspell it "parapsoriasis en plaque."

[e] These terms are frequently used by different authors to denote completely different diseases. For this reason, their use is not recommended.

[f] Poikiloderma vasculare atrophicans is sometimes labeled poikiloderma atrophicans vasculare. Either term is acceptable.

[g] When equivalent terms in different languages are in common use, only the Latin name is given: e.g., parapsoriasis guttata (guttate parapsoriasis, parapsoriasis en gouttes, or en gotas); parapsoriasis lichenoides (lichenoid parapsoriasis, parapsoriasis lichenoides); parapsoriasis variegata (variegate parapsoriasis).

[h] Some authorities regard retiform parapsoriasis as a separate entity.

[i] Some authors regard digitate dermatosis, of which fingerprint parapsoriasis is a synonym and xanthoerythrodermia perstans a variant, as a separate entity.

[j] Xanthoerythrodermia perstans is a variant of digitate dermatosis in which the lesions are yellow in color and on clinical examination. It sometimes incorrectly applied to other forms of parapsoriasis with yellow lesions.

[k] Xanthoerythrodermia perstans is an unnecessary term that should be avoided.

[l] Some authors consider acute and chronic pityriasis lichenoides as separate entities.

CUTANEOUS FINDINGS IN PRIMARY IMMUNODEFICIENCY DISORDERS										
	S. aureus infections									
Disorder	Superficial pyodermas	Abscesses	CMC	Warts	Eczematous dermatitis	Granulomatous dermatitis* (non-infectious)	LE	SVV	Ulcers (PG-like)	Other findings
Ataxia–telangiectasia	+	+			+	+ (often ulcerate)				Oculocutaneous telangiectasias, progeric changes, CALM, BCC
Chédiak–Higashi syndrome	+	+							+	Pigmentary dilution, hyperpigmentation in sun-exposed sites, silvery hair, bleeding diathesis, gingivitis
Chronic granulomatous disease	++	++	+		+	++ (nodular, necrotic)	+		+	DLE in female carriers, Sweet syndrome, oral ulcers
Chronic mucocutaneous candidiasis			++			(++, but infectious due to *Candida*)				Dermatophyte infections, vitiligo, alopecia areata
Common variable immunodeficiency	+	+	+	+	+	++	+	+	+	Dermatophyte infections, vitiligo, alopecia areata
Complement deficiencies	+	+	+				++	+	+	Dermatomyositis, urticaria, lipodystrophy (C3), JIA
DiGeorge syndrome†			+		+	+				
Hyper-IgE syndromes (HIES)	++	++ (cold)	+	‡	++					Neonatal papulopustular eruption
Hyper-IgM syndrome	+	+		+		+	+		+	Oral ulcers
Idiopathic $CD4^+$ lymphocytopenia			+	++			+			$CD4^+$ T cells <300/mm^3 or <20%^, severe HSV infections, cryptococcal meningitis, non-tuberculous mycobacterioses, lymphoma
IgA deficiency	+	+	+		+		+	+	+	Vitiligo, lipodystrophia centrifugalis abdominalis
IgM deficiency	+	+		+	+		+			
IL-1 receptor-associated kinase-4 (IRAK-4) deficiency	++	++ (cold)	+							
Leukocyte adhesion deficiency		++ (necrotic)							++	Poor wound healing, delayed separation of the umbilical stump, gingivitis
SCID	+	+	+	+	+	+*				GVHD, erythroderma; Omenn syndrome
TAP deficiency						++		+	+	
WHIM syndrome	+	+		++						
Wiskott–Aldrich syndrome	++	+			++	+		+		Bleeding diathesis
X-linked agammaglobulinemia	+	++			+	+				Dermatomyositis-like eruption (due to echovirus); ecthyma gangrenosum

*Extensive cutaneous and extracutaneous granulomatous disease (including destructive midfacial granulomas) has been described in children with hypomorphic *RAG1* or *RAG2* mutations.
†Defective thymic function, hypocalcemia secondary to hypoparathyroidism, congenital heart defects, and craniofacial anomalies; due to 22q11 deletions.
‡Patients with AR-HIES due to DOCK8 deficiency are also at increased risk of developing mucocutaneous squamous cell carcinoma and severe warts, molluscum contagiosum, and herpes simplex or varicella–zoster viral infections.
^On more than one occasion with no evidence of HIV infection and the absence of any defined immunodeficiency or immunosuppressive therapy.

Table 60.1 Cutaneous findings in primary immunodeficiency disorders. +, occasional finding; ++, common finding; BCC, basal cell carcinoma; CALM, café-au-lait macules; CMC, chronic mucocutaneous candidiasis; DLE, discoid lupus erythematosus; GVHD, graft-versus-host disease; HSV, herpes simplex virus; IL, interleukin; JIA, juvenile idiopathic arthritis; LE, lupus erythematosus; PG, pyoderma gangrenosum; SCID, severe combined immunodeficiency; SVV, small vessel vasculitis; TAP, transporter associated with antigen processing; WHIM, warts, hypogammaglobulinemia, infections and myelokathexis.

hyper- and/or hypopigmentation, with chromosomal instability likely explaining their predisposition to pigmentary mosaicism. Other dermatologic findings that have been described in individuals with AT include a facial papulosquamous rash, poikiloderma, hypertrichosis favoring the arms, warts, and acanthosis nigricans[12].

Cerebellar ataxia usually begins during infancy and is characterized by swaying of the head and trunk. Choreoathetosis, dysarthric speech, oculomotor abnormalities, and myoclonic jerks often become prominent during childhood. Despite good muscle strength, patients are typically confined to a wheelchair by 11 years of age. The facies become

COMPLEMENT DISORDERS		
Complement component	**Common infectious agents**	**Autoimmune/inflammatory disorders**
Classical pathway		
C1q	Encapsulated bacteria, *Candida* spp.	SLE*, dermatomyositis, GN
C1r	Encapsulated bacteria	SLE*, GN
C1s	Encapsulated bacteria	SLE*
C1 inhibitor		Hereditary angioedema ≫ SLE, GN, partial lipodystrophy, vasculitis
C4	Encapsulated bacteria	SLE* with PPK and scars, HSP, GN, urticaria, JIA‡
C2†	Encapsulated bacteria, especially *Streptococcus pneumoniae*	SLE*, SCLE, DLE, dermatomyositis, HSP, other vasculitides, atrophoderma, cold urticaria, JIA‡, IBD, atherosclerosis
Lectin pathway		
MBL§	Encapsulated bacteria, especially *N. meningitides*	SLE, dermatomyositis, atherosclerosis, chronic pulmonary disease
MASP-2	Encapsulated bacteria, especially *S. pneumoniae*	SLE, IBD
C3 and alternative pathway		
C3	Encapsulated bacteria (severe infections)	SLE, vasculitis, lipodystrophy (partial [cephalo]thoracic), GN, "Leiner phenotype"
Factor H	Encapsulated bacteria	SLE, HUS, GN, age-related macular degeneration
Factor I (C3b inactivator)	Encapsulated bacteria	SLE, HUS, aquagenic urticaria, angioedema
Properdin (XLR)	*Neisseria* spp. (fulminant infections)	
Factor D	*Neisseria* spp.	
Membrane attack complex		
C5 dysfunction	Gram-negative bacteria	"Leiner phenotype"
C5 deficiency	*Neisseria* spp., *S. pneumoniae*	SLE
C6	*Neisseria, Brucella, Toxoplasma* spp.	SLE, JIA, GN
C7	*Neisseria* spp.	SLE, limited systemic sclerosis, ankylosing spondylitis
C8α	*Neisseria* spp.	SLE, fever with HSM, eosinophilia, hypergammaglobulinemia
C8β	*Neisseria* spp.	SLE, JIA
C9	*Neisseria* spp.	

*Hierarchy of SLE risk with homozygous mutations: C1q (~90%) > C1r/s > C4 > C2 (~10–20%).
†Most common of the homozygous complement deficiencies.
‡Especially in heterozygous girls.
§Very low penetrance.

Table 60.8 Complement disorders. Unless otherwise specified, affected individuals usually have biallelic defects. Pyogenic infections caused by encapsulated organisms (e.g. *Streptococcus pneumoniae*, *Haemophilus influenzae*, *Streptococcus pyogenes*) in individuals with classical pathway component deficiencies are not as frequent as neisserial infections in those with membrane attack complex component deficiencies. DLE, discoid lupus erythematosus; GN, glomerulonephritis; HSM, hepatosplenomegaly; HSP, Henoch–Schönlein purpura; HUS, hemolytic uremic syndrome; IBD, inflammatory bowel disease; INH, inhibitor; JIA, juvenile idiopathic arthritis; MBL, mannose-binding lectin; MASP, MBL-associated serum protease; PPK, palmoplantar keratoderma; SCLE, subacute cutaneous lupus erythematosus; SLE, systemic lupus erythematosus; XLR, X-linked recessive. *Adapted with permission from Schachner L, Hansen R (eds). Pediatric Dermatology, 4th edn. London: Mosby, 2011.*

Epidemiology

The incidence of CGD is ~1 in 200 000 live births[56,57]. Ninety percent of patients are boys. Approximately three-quarters of cases have X-linked recessive transmission, while the remainder are autosomal recessive.

Pathogenesis

The defects in CGD involve five subunits of the phagocyte NADPH oxidase: the membrane-bound gp91^{phox} (phagocyte oxidase) and p22^{phox}; and the cytoplasmic p47^{phox}, p67^{phox}, and p40^{phox} (Table 60.10). The failure of microbial killing that characterizes CGD results from an inability of the NADPH oxidase system to rapidly generate toxic reactive oxygen species (ROS) by transferring electrons from NADPH to molecular O_2 (the "respiratory burst") after phagocytosis of a pathogen. Regardless of the gene affected in patients with CGD, having residual NADPH oxidase activity and the resultant ability to generate ROS is associated with less severe disease and longer survival[58,59].

NADPH oxidase activity results in the activation of antimicrobial proteases within the phagosome and the formation of microbicidal "neutrophil extracellular traps" (NETs)[60,61]. In addition to killing and degrading microorganisms, ROS produced by the NADPH oxidase system have roles in the regulation of cytokine synthesis as well as induction of neutrophil apoptosis, which prevents tissue damage at sites of inflammation. Animal models of CGD have shown that a lack of ROS leads to excess inflammation via decreased regulatory T-cell activity, unrestrained γ/δ T-cell activity, and augmented production of cytokines such as IL-1β, IL-8, and IL-17. NADPH oxidase also has a role in modulating MHC class II antigen presentation by B cells[62].

Clinical Features

The skin, perianal area, lymph nodes, and lungs are the sites most often affected by CGD. In general, the X-linked form tends to be more severe than the autosomal recessive forms, with a younger mean age at diagnosis (3 years versus 8 years)[56]. Organisms that commonly cause infections in patients with CGD are listed in Table 60.11[63]. Most carriers of CGD do not have an increased susceptibility to infections.

PRIMARY IMMUNOGLOBULIN DEFICIENCY DISORDERS						
Disorder	**Gene**	**Protein (function)**	**Ig levels**	**B cells**	**Infectious and systemic manifestations**	**Cutaneous manifestations**
Block in B-cell differentiation at the pro-B- to pre-B-cell transition						
X-linked agammaglobulinemia	*BTK*	Bruton tyrosine kinase (pre-B-cell receptor [BCR] signaling)	All ↓	↓↓	• Recurrent infections with *Staphylococcus, Streptococcus*, *Pneumococcus*, *Haemophilus* and *Pseudomonas* spp. • Hepatitis B and enteroviral infections • Lymphomas (~5%)	• The skin is the most common site of infection - Furuncles and cellulitis - Ecthyma gangrenosum • Eczematous dermatitis • Papular dermatitis due to lymphohistiocytic infiltration • Non-infectious granulomas • Dermatomyositis-like disorder associated with chronic echoviral meningoencephalitis
AR agammaglobulinemia	*IGHM*	μ heavy chain of IgM (component of pre-BCR)				
	CD79A, CD79B	Igα chain, Igβ chain (bind μ heavy chain)				
	IGLL1	λ5 (surrogate light chain of pre-BCR)				
	BLNK	B-cell linker protein (binds Bruton tyrosine kinase)				
AD agammaglobulinemia	*LRRC8A*	Leucine-rich repeat-containing 8 family member A				
Defective class switch recombination (e.g. from IgM to IgG, IgA or IgE), somatic hypermutation*, and related mechanisms						
Common variable immunodeficiency (CVID)†	*ICOS*	Inducible costimulator on activated T cells (T-cell help for B-cell differentiation)	IgG,A ↓; ± IgM ↓	Nl or ↓	• Bacterial sinopulmonary infections (organisms similar to X-linked agammaglobulinemia) • *Giardia* spp. gastroenteritis • Autoimmune diseases, especially hemolytic anemia and idiopathic thrombocytopenic purpura • Increased risk of cancer (10-fold overall) and lymphoma (400-fold)	• Pyodermas and mucocutaneous candidiasis • Extensive warts and dermatophyte infections • Eczematous dermatitis • Non-infectious granulomas (Fig. 60.11), which may also involve the lungs, liver and spleen • Autoimmune conditions such as vitiligo, alopecia areata and vasculitis • Clonal $CD8^+$ lymphocytic infiltration of the skin • Clinical manifestations of LRBA deficiency variable; include an IPEX-like presentation (see text) and autoimmune lymphoproliferative syndrome (see Table 60.6)
	TNFRSF13B (AD or AR)	Transmembrane activator and CAML interactor (TACI; B-cell isotype switching)				
	TNFRSF13C	B-cell activating factor receptor (BAFFR; B-cell isotype switching)				
	CD19‡	CD19 antigen (B-cell survival and differentiation)				
	MSH5 (AD)	Mismatch repair protein (regulates class switch recombination)				
	LRBA (AR)	LPS-responsive beige-like anchor protein				
Selective IgA deficiency	*TNFRSF13B* (AD)	See CVID section above	IgA ↓§; anti-IgA antibodies in ~half	Nl	• Clinical manifestations in 10–15% • Similar to CVID	• Mucocutaneous candidiasis • Eczematous dermatitis • Autoimmune conditions such as SLE, vitiligo and lipodystrophia centrifugalis abdominalis
	MSH5 (AD)					
Selective IgM deficiency	?	?	IgM ↓	Nl	• Recurrent bacterial infections • Autoimmune diseases	• Extensive warts • Eczematous dermatitis • SLE
TWEAK deficiency	*TWEAK*	TNF-like weak inducer of apoptosis	IgM, A ↓	Nl	• Pneumonia, other bacterial infections • Thrombocytopenia, neutropenia	• Warts

*T-cell priming may also be defective, particularly in CD40–CD40L disorders (leading some authors to classify the latter as subtypes of severe combined immunodeficiency).
†Relatively common, heterogeneous group of disorders characterized by decreased levels of at least two classes of immunoglobulins (IgG, IgA > IgM) and variable defects in T-cell function; currently, ≤10% of patients have an identifiable disease-causing mutation; several additional genes have also been associated with CVID[104].
‡*CD81* is required for CD19 expression, and a homozygous mutation in the CD81 gene was reported in a patient with hypogammaglobulinemia and autoimmune vasculitis.
§Symptomatic patients often develop the immunodeficiency pattern of CVID.

Table 60.15 Primary immunoglobulin deficiency disorders. Conditions are autosomal recessive (AR) unless otherwise specified. AD, autosomal dominant; CAML, calcium-modulating cyclophilin ligand; Ig, immunoglobulin; Nl, normal; SLE, systemic lupus erythematosus.

PRIMARY IMMUNOGLOBULIN DEFICIENCY DISORDERS						
Disorder	**Gene**	**Protein (function)**	**Ig levels**	**B cells**	**Infectious and systemic manifestations**	**Cutaneous manifestations**
Block in B-cell differentiation at the pro-B- to pre-B-cell transition						
Activated PI3Kδ syndrome	*PIK3CD, PIK3R1*	Phosphoinositide-3-kinase catalytic subunit δ (gain-of-function) or regulatory subunit 1 (loss-of-function)	IgG/G_2, A ↓; IgM ↑; also often CD4 lymphopenia	NI or ↓	• Recurrent sinopulmonary infections • Chronic EBV and CMV infections • LAN, HSM • Lymphoma • Autoimmune diseases, e.g. cytopenias • Growth retardation (*PIK3R1*)	• Staphylococcal abscesses, cellulitis • Severe/recurrent HSV and VZV infections, warts, molluscum contagiosum
X-linked hyper-IgM syndrome	*CD40LG*	CD40 ligand (on T cells)	IgM ↑; isohemagglutinins ↑; IgA,E,G ↓↓	NI	• Recurrent sinopulmonary and GI infections with pyogenic bacteria and opportunistic organisms (e.g. PCP) • Neutropenia • Small lymph nodes • Autoimmune diseases, especially thyroiditis and hemolytic anemia	• Pyodermas • Extensive warts • Oral and anogenital ulcers • Non-infectious granulomas • Autoimmune conditions such as SLE
AR hyper-IgM syndromes	*CD40*	CD40 (on B cells)				
	AICDA	Activation-induced cytidine deaminase			• As above, but massive HSM/LAN (with germinal centers) and no opportunistic infections	• Pyodermas
	UNG	Uracil-DNA glycosylase				
Hypohidrotic ectodermal dysplasia with immunodeficiency	*IKBKG (NEMO)* (X-linked recessive)	NF-κB essential modulator (activates NF-κB, which is involved in CD40 signaling)	IgM ↑; ± IgA ↑ ; ± IgG ↓	NI	• Pyogenic bacterial and opportunistic infections • Subset of *NEMO* patients with osteopetrosis and lymphedema	• Pyodermas • Ectodermal dysplasia (see Ch. 63)
	IKBA (*NFKBIA*; AD, gain of function)	Inhibitor of κB-α (inhibits NF-κB)				
Abnormal DNA methylation leading to defective B-cell negative selection and terminal differentiation						
*I*mmunodeficiency, centromeric instability, and *f*acial anomalies (ICF) syndrome	*DNMT3B, ZBTB24*	DNA methyltransferase 3β, zinc finger and BTB-containing 24	All ↓	NI or ↓	• Bacterial sinopulmonary and GI infections • Abnormal facies, intellectual disability	• Telangiectasias (uncommon) • Nevoid hyperpigmentation
Aberrant chemokine signaling						
WHIM syndrome¶	*CXCR4* (AD, gain-of-function)	CXC chemokine receptor 4 (binds CXCL12; key role in bone marrow homeostasis and lymphocyte trafficking)	IgG ↓; ± IgA,M ↓	↓	• Recurrent bacterial sinopulmonary infections • Myelokathexis (mature neutrophils fail to exit the bone marrow)	• Cellulitis and pyodermas • Extensive verruca vulgaris and condyloma acuminata (Fig. 60.12)
Delayed maturation of helper T-cell function						
Transient hypogammaglobulinemia of infancy	?	?	IgG,A ↓ (resolves by age 3 y)	NI	• Failure to thrive in infancy • Recurrent sinopulmonary and GI infections	• Recurrent pyodermas and abscesses • Eczematous dermatitis

¶Warts, hypogammaglobulinemia, infections and myelokathexis; in a phase 1 trial, treatment with plerixafor (AMD 3100), a CXCR4 antagonist, led to higher neutrophil counts and improvement of warts.

Table 60.15 Primary immunoglobulin deficiency disorders. (cont'd) Conditions are autosomal recessive (AR) unless otherwise specified. AD, autosomal dominant; GI, gastrointestinal; HSM, hepatosplenomegaly; HSV, herpes simplex virus; Ig, immunoglobulin; *IKBKG*, inhibitor of k light polypeptide gene enhancer in B cells, kinase g; LAN, lymphadenopathy; NI, normal; PCP, *Pneumocystis jiroveci* pneumonia; SLE, systemic lupus erythematosus; VZV, varicella–zoster virus.

well as Gram-negative bacteria. Intravenous therapy should be continued for at least 10–14 days, followed by a several-week course of oral antibiotics. Surgical interventions such as debridement, irrigation, and prolonged drainage are important for deeper infections.

Chronic prophylaxis with trimethoprim–sulfamethoxazole has been shown to decrease the incidence of bacterial infection in CGD patients without increasing the incidence of fungal infections. The rate of *Aspergillus* spp. infections has been reduced by prophylaxis with itraconazole[70]. Patients with CGD have shown clinical improvement after administration of interferon-γ, which likely augments oxidant-independent antimicrobial pathways. Granulocyte transfusions have been used for rapidly progressive, life-threatening infections. Short courses of systemic corticosteroids have also been helpful for patients with obstructive granulomas of the bronchopulmonary, gastrointestinal, or genitourinary tract[53]. Additional immunomodulatory therapies with potential benefit for inflammatory manifestations of CGD include azathioprine, hydroxychloroquine, anakinra, thalidomide, pioglitazone (increases ROS production and efferocytosis), and possibly sirolimus[71,72]; tumor necrosis factor (TNF) inhibitors may improve colitis but increase the risk of infectious complications.

Hematopoietic stem cell transplantation represents a potentially curative treatment for CGD. Although younger patients without infection at the time of transplantation have the best outcomes (survival >95%), use of reduced-intensity conditioning regimens has enabled successful transplantation of higher-risk patients such as adults and individuals with recalcitrant infections or inflammatory manifestations[73]. Children with CGD who undergo transplantation have improved growth and fewer infections, surgical interventions, and hospital admissions than those managed conservatively[74].

Gene therapy was initially performed in five adults with the $p47^{phox}$-deficient form of CGD, and a single infusion of transduced $CD34^+$

OTHER INHERITED PHAGOCYTE DEFECTS

Disorder	Inh	Gene	Protein (defect or function)	Clinical features
Selected disorders featuring neutropenia or neutrophil defects				
Severe congenital neutropenia	AD	*ELANE*	Neutrophil elastase (abnormal elastase trafficking and accumulation)	• Neutropenia • MDS, AML
	AD	*GFI1*	Transcriptional repressor of elastase (abnormal elastase accumulation)	• Neutropenia, lymphopenia • Circulating myeloid progenitors
	AR	*HAX1*	Mitochondrial HS1-associated protein X1 (protects against apoptosis in myeloid cells)	• Neutropenia • Increased apoptosis of myeloid cells
	AD	*CSF3R*	Granulocyte colony-stimulating factor receptor	• Neutropenia • Severe myeloid hypoplasia
	AR	*G6PC3*	Glucose 6 phosphatase catalytic subunit 3 (glucose metabolism)	• Neutropenia, thrombocytopenia • Urogenital and cardiac malformations • Ectatic veins on trunk/extremities
Cyclic neutropenia	AD	*ELANE*	Neutrophil elastase	• Alternating 21-day cycles of neutropenia & monocytopenia • Fever and oral ulcers at nadir
X-linked neutropenia	XR	*WASP*	WASP (gain of function; see text, Wiskott–Aldrich syndrome)	• Neutropenia
p14 deficiency	AR	*LAMTOR2*	Endosomal adaptor protein p14 (p14; endosomal biogenesis)	• Neutropenia • Pneumococcal infections • Diffuse pigmentary dilution of the skin and hair • Short stature, coarse facies
Shwachman–Bodian–Diamond syndrome	AR	*SBDS*	SBDS protein (ribosomal RNA metabolism)	• Neutropenia > pancytopenia • MDS, AML • Exocrine pancreatic insufficiency • Chondrodysplasia
Poikiloderma with neutropenia, Clericuzio type	AR	*USB1*	U6 snRNA biogenesis phosphodiesterase 1	• Neutropenia • Dermatitis → poikiloderma, keratotic papules • Recurrent skin & respiratory infections
Specific granule deficiency	AR	*CEBPE*	C/EBPε transcription factor (granulocyte differentiation)	• Bilobed neutrophils • Recurrent bacterial infections
Myeloperoxidase deficiency	AR	*MPO*	Myeloperoxidase (microbial killing by granulocytes)	• *Candida* spp. and *S. aureus* infections • Often asymptomatic
Leukocyte adhesion deficiencies (LADs) and related conditions				
LAD-I	AR	*ITGB2*	β_2-integrin subunit of LFA-1, CR3, and p150 (see text)	• Neutrophilia; ↓ tissue neutrophils • Necrotic abscesses, ulcers • Poor wound healing, delayed umbilical stump separation • Gingivitis • Bleeding diathesis (LAD-III)
LAD-II	AR	*SLC35C1 (FUCT1)*	GDP-fucose transporter 1 (sialyl-Lewis X expression; see text)	
LAD-III	AR	*FERMT3*	Kindlin-3 (defective integrin activation; see text)	
Rac2 deficiency	AD	*RAC2*	Rac2 GTPase (NADPH oxidase, integrin-dependent adhesion, and neutrophil migration; see Table 60.10)	

Table 60.13 Other inherited phagocyte defects. See text and Tables 60.5 and 60.10 for Chédiak–Higashi syndrome and chronic granulomatous disease. AD, autosomal dominant; AML, acute myeloid leukemia; AR, autosomal recessive; BCG, bacillus Calmette–Guérin; IFN, interferon; IL, interleukin; Inh, inheritance; LFA-1, lymphocyte function-associated antigen-1; MDS, myelodysplastic syndrome; WASP, Wiskott–Aldrich syndrome protein; XR, X-linked recessive.

OTHER INHERITED PHAGOCYTE DEFECTS				
Disorder	**Inh**	**Gene**	**Protein (defect or function)**	**Clinical features**
Defects resulting in predisposition to specific infections				
Defects of the interleukin-12/ interferon-γ axis	AR	*IL12B*	Subunit of IL-12 and IL-23 (stimulation of IFN-γ production)	• Severe mycobacterial and *Salmonella* infections • Disseminated BCG infection • Chronic mucocutaneous candidiasis (*RORC*, *IL12RB1* > *IL12B*) • Viral infections (*STAT1, TYK2*) • Hyper-IgE syndrome (*TYK2*; see text)
	AR	*IL12RB1*	IL-12 and IL-23 receptor β1 chain	
	AR, AD	*IFNGR1*	IFN-γ receptor (ligand binding)	
	AR	*IFNGR2*	IFN-γ receptor (signaling)	
	AR, AD	*STAT1**	Signal transducer and activator of transcription 1 (STAT1)* (IFN receptor signaling is impaired by loss-of-function mutations)	
	AR	*TYK2*	Tyrosine kinase 2 (IFN/IL-12/other cytokine receptor signaling)	
	AD	*IFR8*	IFN regulatory factor 8 (IL-12 signaling)	
	AR	*ISG15*	IFN-α/β-inducible ubiquitin-like modifier	
	AR	*RORC*	Retinoic acid receptor-related orphan receptor C (IFN-γ and IL-17 signaling)	
Defects in Toll-like receptor (TLR) signaling	AD	*TLR3*	TLR3 (signals IFN-α/β production)	• Herpes simplex encephalitis
	AR	*UNC93B1*	UNC-93B (endoplasmic reticulum protein required for TLR3 signaling)	
	AD	*TRAF3*	TNF receptor-associated protein 3 (TLR3 signaling)	
	AD, AR	*TICAM1* (*TRIF*)	TLR-adaptor molecule 1 (TLR3/4 signaling)	
	AD	*TBK1*	TANK-binding kinase 1 (TLR3 signaling)	
	AR	*IRAK4*	IL-1 receptor associated kinase-4 (IL-1 receptor & TLR signaling)	• Recurrent pyogenic sinopulmonary and skin infections with *Streptococcus pneumoniae* and *S. aureus*, respectively
	AR	*MYD88*	Myeloid differentiation primary response gene 88 (recruits IRAK4 to the IL-1 receptor and TLRs)	
GATA2 deficiency/ MonoMAC syndrome	AD	*GATA2*	GATA binding protein 2	• Monocytopenia, B/NK lymphopenia, myelodysplasia, myeloid leukemia • Mycobacterial and fungal infections (e.g. histoplasmosis) • Recalcitrant warts • Lymphedema • Alveolar proteinosis

*Gain-of-function *STAT1* mutations lead to chronic mucocutaneous candidiasis (see Table 60.4) and/or confer susceptibility to endemic dimorphic fungal infections; *loss*-of-function *STAT3* mutations underlie AD hyper-IgE syndrome (see text) while *gain*-of-function *STAT3* mutations can result in lymphoproliferation, autoimmunity, hypogammaglobulinemia, mycobacterial infections, and occasionally psoriasiform dermatitis; defects in STAT5B, which is involved in IL-2 and growth hormone receptor signaling, lead to an AR syndrome of growth hormone insensitivity, decreased $CD4^+/CD25^+$ regulatory T cells, viral infections, and eczematous dermatitis.

Table 60.13 Other inherited phagocyte defects. (cont'd) AD, autosomal dominant; AR, autosomal recessive; IFN, interferon; IL, interleukin; Inh, inheritance.

peripheral blood stem cells led to peak levels of corrected granulocytes in 3–6 weeks with persistence for as long as 6 months[75]. Subsequently, two young men with X-linked recessive CGD were treated with non-myeloablative conditioning prior to the infusion of $CD34^+$ peripheral blood stem cells transduced *ex vivo* with a retroviral vector expressing $gp91^{phox}$; this led to sustained engraftment of functionally corrected phagocytes. However, following initial resolution of infections, the transgene was silenced due to methylation of the viral promoter in both patients, and within 3 years they developed myelodysplasia with monosomy 7 as a result of insertional activation of ecotropic viral integration site 1 (*EVI1*)[76]. To increase the safety and efficacy of gene therapy for CGD, current investigations are utilizing approaches such as self-inactivating lentiviral vectors, targeted integration of transgenes into a genomic "safe harbor" site, or the CRISPR-Cas9 site-specific nuclease system to encourage repair of the endogenous gene via homologous recombination (see Ch. 3)[77,78].

Dihydrorhodamine 123 flow cytometric studies may be utilized in determining the carrier status of the sisters and other female relatives of patients with X-linked CGD, which is important for genetic counseling prior to pregnancy. Prenatal diagnosis of CGD is also possible.

HYPERIMMUNOGLOBULIN E SYNDROMES

Synonyms: ■ Hyper-IgE syndromes ■ Job syndrome ■ Buckley syndrome

Key features

- Characterized by recurrent skin and sinopulmonary tract infections, early-onset eczematous dermatitis, and extremely elevated IgE levels
- The classic form is an autosomal dominant disorder with skin infections that are usually caused by *S. aureus* (in particular "cold abscesses") and, especially during infancy, *C. albicans*
- The autosomal dominant form also features a neonatal papulopustular eruption, progressively coarse facies, osteopenia with fractures, scoliosis, and dental abnormalities
- Autosomal recessive forms have a broader spectrum of infectious complications

CUTANEOUS FINDINGS IN PRIMARY IMMUNODEFICIENCY DISORDERS										
	S. aureus infections									
Disorder	Superficial pyodermas	Abscesses	CMC	Warts	Eczematous dermatitis	Granulomatous dermatitis* (non-infectious)	LE	SVV	Ulcers (PG-like)	Other findings
Ataxia–telangiectasia	+	+			+	+ (often ulcerate)				Oculocutaneous telangiectasias, progeric changes, CALM, BCC
Chédiak–Higashi syndrome	+	+							+	Pigmentary dilution, hyperpigmentation in sun-exposed sites, silvery hair, bleeding diathesis, gingivitis
Chronic granulomatous disease	++	++	+		+	++ (nodular, necrotic)	+		+	DLE in female carriers, Sweet syndrome, oral ulcers
Chronic mucocutaneous candidiasis			++			(++, but infectious due to *Candida*)				Dermatophyte infections, vitiligo, alopecia areata
Common variable immunodeficiency	+	+	+	+	+	++	+	+	+	Dermatophyte infections, vitiligo, alopecia areata
Complement deficiencies	+	+	+				++	+	+	Dermatomyositis, urticaria, lipodystrophy (C3), JIA
DiGeorge syndrome†			+		+	+				
Hyper-IgE syndromes (HIES)	++	++ (cold)	+	‡	++					Neonatal papulopustular eruption
Hyper-IgM syndrome	+	+		+		+	+		+	Oral ulcers
Idiopathic $CD4^+$ lymphocytopenia			+	++			+			$CD4^+$ T cells <300/mm^3 or <20%^, severe HSV infections, cryptococcal meningitis, non-tuberculous mycobacterioses, lymphoma
IgA deficiency	+	+	+		+		+	+	+	Vitiligo, lipodystrophia centrifugalis abdominalis
IgM deficiency	+	+		+	+		+			
IL-1 receptor-associated kinase-4 (IRAK-4) deficiency	++	++ (cold)	+							
Leukocyte adhesion deficiency		++ (necrotic)							++	Poor wound healing, delayed separation of the umbilical stump, gingivitis
SCID	+	+	+	+	+	+*				GVHD, erythroderma; Omenn syndrome
TAP deficiency						++		+	+	
WHIM syndrome	+	+		++						
Wiskott–Aldrich syndrome	++	+			++	+		+		Bleeding diathesis
X-linked agammaglobulinemia	+	++			+	+				Dermatomyositis-like eruption (due to echovirus); ecthyma gangrenosum

*Extensive cutaneous and extracutaneous granulomatous disease (including destructive midfacial granulomas) has been described in children with hypomorphic *RAG1* or *RAG2* mutations.
†Defective thymic function, hypocalcemia secondary to hypoparathyroidism, congenital heart defects, and craniofacial anomalies; due to 22q11 deletions.
‡Patients with AR-HIES due to DOCK8 deficiency are also at increased risk of developing mucocutaneous squamous cell carcinoma and severe warts, molluscum contagiosum, and herpes simplex or varicella–zoster viral infections.
^On more than one occasion with no evidence of HIV infection and the absence of any defined immunodeficiency or immunosuppressive therapy.

Table 60.1 Cutaneous findings in primary immunodeficiency disorders. +, occasional finding; ++, common finding; BCC, basal cell carcinoma; CALM, café-au-lait macules; CMC, chronic mucocutaneous candidiasis; DLE, discoid lupus erythematosus; GVHD, graft-versus-host disease; HSV, herpes simplex virus; IL, interleukin; JIA, juvenile idiopathic arthritis; LE, lupus erythematosus; PG, pyoderma gangrenosum; SCID, severe combined immunodeficiency; SVV, small vessel vasculitis; TAP, transporter associated with antigen processing; WHIM, warts, hypogammaglobulinemia, infections and myelokathexis.

hyper- and/or hypopigmentation, with chromosomal instability likely explaining their predisposition to pigmentary mosaicism. Other dermatologic findings that have been described in individuals with AT include a facial papulosquamous rash, poikiloderma, hypertrichosis favoring the arms, warts, and acanthosis nigricans[12].

Cerebellar ataxia usually begins during infancy and is characterized by swaying of the head and trunk. Choreoathetosis, dysarthric speech, oculomotor abnormalities, and myoclonic jerks often become prominent during childhood. Despite good muscle strength, patients are typically confined to a wheelchair by 11 years of age. The facies become

INHERITED DISORDERS CHARACTERIZED BY LYMPHOPROLIFERATION DUE TO IMMUNE DYSREGULATION				
Disorder	**Inh**	**Gene**	**Protein (function)**	**Clinical features**
Chédiak–Higashi syndrome	AR	*LYST*	See Table 60.5	See Table 60.5
Griscelli syndrome type 2	AR	*RAB27A*		
Familial hemophagocytic lymphohistiocytosis	AR	*PRF1*	Perforin (major cytolytic granule protein of CTL and NK cells)	• Hemophagocytic syndrome • Absent NK cell cytotoxicity • Fatal during early childhood unless hematopoietic stem cell transplantation is performed
	AR	*UNC13D*	Unc13 homolog D (primes cytolytic granules for secretion)	
	AR	*STX11*	Syntaxin 11 (involved in cytolytic granule trafficking and fusion)	
	AR	*STXBP2*	Syntaxin-binding protein 2 (regulates cytolytic granule trafficking)	
X-linked lymphoproliferative syndrome (Duncan disease)	XR	*SH2D1A*	Signaling lymphocyte activation molecule-associated protein (adaptor protein that regulates B-, T- and NK-cell function; particularly important for immune responses to EBV)	• Severe EBV-related infectious mononucleosis in childhood or adolescence (e.g. fever, pharyngitis, LAN, HSM), often associated with morbilliform eruptions, purpura and/or jaundice • ± Hypogammaglobulinemia • Fatal EBV-related B-cell lymphoma (70%) • Lymphocytic vasculitis
	XR	*XIAP*	X-linked inhibitor of apoptosis	
EBV-associated lymphoproliferative syndrome	AR	*ITK*	IL-2-inducible T-cell kinase	
XMEN (*X*-linked immunodeficiency with *m*agnesium defect, *E*BV infection, and *n*eoplasia)	XR	*MAGT1*	Magnesium transporter 1	• Decreased $CD4^+$ T cells • EBV infection with persistent viremia; recurrent sinopulmonary infections • EBV-associated lymphoma
STK4 deficiency	AR	*STK4*	Serine/threonine kinase 4	• EBV-associated lymphoproliferation and lymphoma • Recurrent bacterial, viral (including warts, molluscum, HSV), and candidal infections • Autoimmune cytopenias
Autoimmune lymphoproliferative syndrome (Canale–Smith syndrome) and related conditions*	AD >AR	*TNFRSF6*	CD95 (Fas; cell-surface apoptosis receptor)	• Massive LAN and/or splenomegaly • Increased $CD4^-/CD8^-$ α/β T cells • Autoimmune cytopenias, LE, small vessel vasculitis • Recurrent bacterial and viral infections (*CASP8*) • Increased risk of lymphoma
	AD >AR	*TNFSF6*	CD95L (Fas ligand)	
	AR	*FADD*	Fas-associated death domain	
	AD > AR	*CASP10*	Caspase 8 and 10 (proteases in intracellular apoptosis cascade)	
	AR	*CASP8*		
	Mosaic	*NRAS*, *KRAS*	NRAS, KRAS (gain of function leads to decreased lymphocyte apoptosis)	
	AR	*PRKCD*	Protein kinase C δ (regulates B-cell proliferation)	
	AD	*CTLA4*	Cytotoxic T lymphocyte-associated protein 4 (costimulation of T cells)	
Immunodeficiency with lymphoid proliferation	AR	*IL2RA*	IL-2 receptor α-chain (apoptosis of developing T cells in the thymus)	• Bacterial, viral, and fungal infections • Extensive lymphocytic infiltration of the liver, lung, gut and bones • Eczematous dermatitis; autoimmune polyendocrinopathy, cytopenias and enteropathy

**LRBA* mutations (see Table 60.15) and gain-of-function *CARD11* or *STAT3* mutations can also lead to autoimmune lymphoproliferative syndrome-like disorders.

Table 60.6 Inherited disorders characterized by lymphoproliferation due to immune dysregulation. Hemophagocytic syndromes result from uncontrolled T-cell and macrophage activation, manifesting with fever, pancytopenia, hepatosplenomegaly (HSM) and lymphadenopathy (LAN) (due to lymphohistiocytic infiltration), hypertriglyceridemia, and hypofibrinogenemia. AD, autosomal dominant; AR, autosomal recessive; CTL, cytotoxic T lymphocytes; Inh, inheritance; LE, lupus erythematosus; NK, natural killer; XR, X-linked recessive.

COMPLEMENT DISORDERS

Key features

- Deficiency or dysfunction of early complement components increases susceptibility to pyogenic infections caused by encapsulated bacteria and autoimmune disorders, especially systemic lupus erythematosus
- Deficiency of late complement components leads to a markedly increased risk of neisserial infections

Introduction

The complement system represents an important effector of the innate immune response (see Ch. 4). An enzymatic cascade of complement activation can be triggered by three different pathways: classical, alternative, and lectin (Fig. 60.6). In addition to their roles in killing microbial pathogens, complement proteins serve as regulators of a variety of humoral and cellular immune functions (Table 60.7). As a result, clinical manifestations of complement deficiency include autoimmune diseases as well as increased susceptibility to infection[49,50].

TYPES OF SEVERE COMBINED IMMUNODEFICIENCY (SCID)			
Disorder	**Gene**	**Protein function and specific clinical features**	**Cells**
Defects in cytokine receptors and their signaling			
X-linked SCID	*IL2RG*	Common γ chain (γ_c) of the receptors for IL-2, 4, 7, 9, 15, and 21	$T^-/B^+/NK^-$
JAK3 deficiency	*JAK3*	Tyrosine kinase with a role in signal transduction via the receptors for IL-2, 4, 7, 9, 15, and 21	$T^-/B^+/NK^-$
Interleukin-7 receptor deficiency	*IL7R*	α-chain of the IL-7 receptor; critical for development and differentiation of T cells; possible Omenn syndrome phenotype	$T^-/B^+/NK^+$
Increased apoptosis of lymphocytes ± other hematopoietic cells			
Defects in purine pathway enzymes that result in the build-up of toxic metabolites			
Adenosine deaminase (ADA) deficiency	*ADA*	Enzyme in the purine salvage pathway	$T^-/B^-/NK^-$
Purine nucleoside phosphorylase (PNP) deficiency	*PNP*	Enzyme in the purine salvage pathway (downstream of ADA)	$T^-/B^-/NK^-$
Defects disrupting mitochondrial energy production			
Reticular dysgenesis (aleukocytosis)	*AK2*	Adenylate kinase 2, a mitochondrial enzyme that regulates ADP levels in leukocytes; profound neutropenia and sensorineural deafness	$T^-/B^-/NK^-$
Defects disrupting actin polymerization			
Coronin 1A deficiency	*CORO1A*	Actin regulator expressed in T-cell precursors; severe warts, molluscum contagiosum, and herpesvirus infections	$T^-/B^+/NK^+$
Defects in (pre-)T-cell receptors (TCRs) ± B-cell receptors (BCRs) and their signaling			
Omenn syndrome	*RAG1* and *RAG2*	Mediate V(D)J recombination of the genes that encode BCRs (immunoglobulins) and TCRs§; erythroderma, alopecia, LAN, HSM, diarrhea, eosinophilia, elevated IgE levels (see text)	$T^-/B^-/NK^+$
Artemis deficiency	*DCLRE1C (Artemis)*	Repairs the DNA double-stranded breaks that occur during V(D)J recombination and from ionizing radiation§; possible Omenn syndrome phenotype	$T^-/B^-/NK^+$
PRKDC deficiency	*PRKDC*	DNA-activated protein kinase catalytic subunit critical for V(D)J recombination§	$T^-/B^-/NK^+$
LIG4 syndrome	*LIG4*	Ligase component of the DNA non-homologous end-joining (NHEJ) machinery required for the repair of DNA double-stranded breaks§; photosensitivity, telangiectasias, bird-like facies, microcephaly	$T^-/B^-/NK^+$
NHEJ1 (Cernunnos) deficiency	*NHEJ1*	NHEJ factor required for the repair of DNA double-stranded breaks§; bird-like facies, microcephaly	$T^-/B^-/NK^+$
T-cell receptor deficiencies	*CD3γ, CD3δ, CD3ε,* and *CD3ζ*	TCR-associated proteins required for cell surface expression of and signal transduction by the TCR; most severe with defective CD3δ	$T^-/B^+/NK^+$
	TRAC	TCR-α constant region; relatively mild immunodeficiency	
CD45 deficiency	*PTPRC*	Transmembrane tyrosine phosphatase with a role in TCR signaling	$T^-/B^+/NK^+$
ZAP70 deficiency	*ZAP70*	Tyrosine kinase with a role in TCR signaling	$CD4^+CD8^-/B^+/NK^+$
CD8 deficiency¶	*CD8A*	Co-receptor for TCRs recognizing MHC class I-associated peptides; role in TCR signaling	$CD4^+CD8^-/B^+/NK^+$
Calcium channel deficiency	*ORAI1*	Pore-forming subunit of calcium channel with a role in TCR signaling; hypohidrotic ectodermal dysplasia and myopathy	$T^+/B^+/NK^+$
	STIM1	Calcium channel component with a role in TCR signaling; LAN, HSM, autoimmune cytopenias, dental dysplasia, myopathy	$T^+/B^+/NK^+$
Antigen presentation defects (bare lymphocyte syndromes)			
MHC class II deficiency	*CIITA, RFXANK, RFX5, and RFXAP*	Transactivate the MHC class II promoter; MHC class II molecules are required for the positive selection of $CD4^+$ T cells in the thymus as well as for antigen presentation to $CD4^+$T cells	$T^+/B^+/NK^+$, but fewer and dysfunctional $CD4^+$ T cells
MHC class I deficiency¶	*TAP1*, *TAP2, TAPBP, B2M*	*T*ransporter associated with *a*ntigen *p*rocessing (TAP) and the TAP binding protein (tapasin); translocate peptides from the cytosol to MHC class I molecules in the ER; cutaneous granulomas in TAP deficiency	$CD4^+CD8^-/B^+/NK^+$
Defective thymic development			
Winged helix nude (forkhead box N1) deficiency	*WHN (FOXN1)*	Transcription factor important to thymic development; complete congenital alopecia and nail dystrophy (human equivalent of "nude" mice)	$T^-/B^+/NK^+$

§Patients have increased sensitivity to ionizing radiation.
¶Not true SCID

Table 60.16 Types of severe combined immunodeficiency (SCID). "Leaky SCID" refers to less severe deficits in T-cell number and function due to hypomorphic (milder) mutations in SCID-associated genes. Cartilage–hair hypoplasia (see text) also occasionally presents as SCID with an Omenn-like phenotype. Two most common forms of SCID, accounting for approximately two-thirds of cases, are shaded. B2M, β2-microglobulin; *DCLRE1C*, DNA cross-link repair protein 1C; ER, endoplasmic reticulum; HSM, hepatosplenomegaly; LAN, lymphadenopathy; MHC, major histocompatibility complex; V(D)J, variable/diversity/joining.

defective function of both cell-mediated and humoral immunity [1–3]. The combined frequency of all forms of SCID is 1 in 30,000 to 1 in 100,000 live births, and approximately 75% of patients are male. Almost half of affected individuals have an X-linked recessive form of SCID, which is usually caused by defects in the gene encoding the common γ (γ_c) chain of the IL-12 receptor. Other forms of SCID have autosomal recessive inheritance and include adenosine deaminase (ADA) deficiency (20% of SCID) and JAK3 deficiency (approximately 5% of SCID).

Pathogenesis. The molecular defects that can lead to a SCID phenotype are summarized in Table 177.8 [1,2].

Clinical features. Patients with SCID typically begin to have recurrent infections, chronic diarrhoea and failure to thrive during the first few months of life. Early infections often include mucocutaneous candidiasis, persistent viral gastroenteritis and pneumonias resulting from bacteria, viruses or *P. jiroveci*. Skin infections are commonly caused by *S. aureus* and *Strep. pyogenes* as well as *Candida* spp. Despite their frequent infections, patients with SCID usually lack tonsillar buds and palpable lymphoid tissue.

Infants with SCID may present with morbilliform or seborrhoeic dermatitis-like eruptions [4,5], which often reflect maternofetal GVHD (see Chapter 178). Cutaneous GVHD in patients with SCID may also resemble lichen planus, acrodermatitis enteropathica, Langerhans' cell histiocytosis, ichthyosiform erythroderma or scleroderma. Skin biopsy specimens show the histopathological characteristics of GVHD. Extensive eczematous dermatitis or severe exfoliative erythroderma, often associated with alopecia, may occur without GVHD in infants with Omenn syndrome. Additional features of Omenn syndrome include lymphadenopathy, hepatosplenomegaly and peripheral eosinophilia.

Most patients with SCID have a profound deficiency of T cells and a low absolute lymphocyte count. Patients are further classified by the results of fluorescent-activated cell sorter analysis into those with and without B and NK cells. Diagnosis of a particular form of SCID can be established by genetic testing, flow cytometric analysis of peripheral blood mononuclear cells with antibodies directed against a particular protein (e.g. γ_c or JAK3) that is missing from the cell surface, or measurement of red blood cell levels of a defective enzyme (e.g. ADA) or purine metabolites. In families with a previously affected sibling, prenatal diagnosis of SCID has been performed by fetal DNA analysis and ADA assays.

Maternal T-cell engraftment is detectable (requiring maternal cells to comprise ≥1% of peripheral blood leucocytes) in approximately 50% of infants with SCID [5]. In the majority of cases, the presence of these maternal T cells is asymptomatic; in 30–40% of patients, mild findings such as morbilliform eruptions, cutaneous erythema

Table 177.8 Severe combined immunodeficiency (SCID)

Disorder	Gene	Protein function ± specific clinical findings	Cells
Defects in cytokine receptors and their signalling			
X-linked SCID	*IL2RG*	Common γ chain (γ_c) of the receptors for IL-2, 4, 7, 9, 15 and 21	$T^-/B^+/NK^-$
JAK3 deficiency	*JAK3*	Tyrosine kinase with a role in signal transduction via the receptors for IL-2, 4, 7, 9, 15 and 21	$T^-/B^+/NK^-$
Interleukin-7 receptor deficiency	*IL7R*	α chain of the IL-7 receptor; critical for development and differentiation of T cells; occasionally Omenn-like phenotype	$T^-/B^+/NK^+$
Increased apoptosis of lymphocytes ± other haematopoietic cells			
Accumulation of toxic metabolites due to purine pathway enzyme defects			
ADA deficiency	*ADA*	Enzyme in the purine salvage pathway; cupping/flaring of the costochondral junction on chest X-rays; occasionally Omenn-like phenotype	$T^-/B^-/NK^-$
PNP deficiency	*PNP*	Enzyme in the purine salvage pathway that acts downstream of ADA	$T^-/B^-/NK^-$
Defects disrupting mitochondrial energy production			
Reticular dysgenesis (aleucocytosis)	*AK2*	Adenylate kinase 2, a mitochondrial enzyme that regulates ADP levels in leucocytes; profound neutropenia and sensorineural deafness	$T^-/B^-/NK^-$
Defects disrupting actin polymerization			
Coronin 1A deficiency	*CORO1A*	Actin regulator expressed in T-cell precursors	$T^-/B^+/NK^+$

Table 177.8 *Continued*

Disorder	Gene	Protein function ± specific clinical findings	Cells
Defects in (pre-) TCRs +/– BCRs and their signalling			
Omenn syndrome	*RAG1* and *RAG2*	Mediate V(D)J recombination of genes that encode BCRs (immunoglobulins) and TCRs; exfoliative erythroderma, hepatosplenomegaly, lymphadenopathy, eosinophilia	$T^-/B^-/NK^+$
Artemis deficiency	*DCLRE1C (Artemis)*	DNA cross-link repair protein that repairs DNA double-stranded breaks that occur during V(D)J recombination and upon exposure to ionizing radiation; sensitivity to ionizing radiation; occasionally Omenn-like phenotype	$T^-/B^-/NK^+$
PRKDC deficiency	*PRKDC*	DNA-activated protein kinase catalytic subunit critical for V(D)J recombination; sensitivity to ionizing radiation	$T^-/B^-/NK^+$
LIG4 syndrome	*LIG4*	Ligase component of DNA non-homologous end-joining machinery required for repair of DNA double-stranded breaks; sensitivity to ionizing radiation; variable severity, occasionally Omenn-like phenotype; microcephaly, bird-like facies, photosensitivity and telangiectasias	$T^-/B^-/NK^+$
NHEJ1 (Cernunnos) syndrome	*NHEJ1*	Non-homologous end-joining factor required for the repair of DNA double-stranded breaks; sensitivity to ionizing radiation; variable severity; microcephaly and bird-like facies	$T^-/B^-/NK^+$
TCR deficiencies	CD3γ, CD3δ, CD3ε and CD3ζ	TCR-associated proteins required for cell-surface expression of and signal transduction by the TCR; the T-cell defect is most severe with CD3δ defects	$T^-/B^+/NK^+$
CD45 deficiency	*PTPRC*	Transmembrane tyrosine phosphatase with a role in TCR signalling	$T^-/B^+/NK^+$
ZAP-70 deficiency	*ZAP-70*	Tyrosine kinase with a role in TCR signalling; lymphocytosis; variable severity and phenotype, occasionally Omenn-like	$CD4^+CD8^-/B^+/NK^+$
CD8 deficiency	*CD8A*	Co-receptor for TCRs recognizing MHC class I-associated peptides; role in TCR signalling; milder than true SCID	$CD4^+CD8^-/B^+/NK^+$
Calcium channel deficiency	*ORAI1*	Pore-forming subunit of calcium channel with a role in TCR signalling; hypohidrotic ectodermal dysplasia and myopathy	$T^+/B^+/NK^+$
	STIM1	Calcium channel component with a role in TCR signalling; hepatosplenomegaly, lymphadenopathy, autoimmune cytopenias, dental dysplasia, myopathy	$T^+/B^+/NK^+$
Antigen presentation defects (bare lymphocyte syndromes)			
MHC class II deficiency	*CIITA* *RFXANK* *RFX5* *RFXAP*	Transactivate the MHC class II promoter; MHC class II molecules are required for the positive selection of $CD4^+$ T cells in the thymus as well as for antigen presentation to $CD4^+$ T cells	$T^+/B^+/NK^+$; fewer/ dysfunctional $CD4^+$ T cells
MHC class I deficiency	*TAP1, TAP2* and *TAPBP*	TAP and TAP binding protein (tapasin); translocate peptides from the cytosol to MHC class I molecules in the endoplasmic reticulum; milder than true SCID; cutaneous granulomas	$CD4^+CD8^-/B^+/NK^+$
Defective thymic development			
Winged helix nude (forkhead box N1) deficiency	*WHN (FOXN1)*	Transcription factor important to thymic development; congenital alopecia and nail dystrophy (human equivalent of 'nude' mice)	$T^-/B^+/NK^+$

Cartilage–hair hypoplasia (see text) occasionally presents as SCID with an Omenn-like phenotype.
ADA, adenosine deaminase; BCR, B-cell receptor; IL, interleukin; MHC, major histocompatibility complex; NK, natural killer; PNP, purine nucleoside phosphorylase; TAP, transporter associated with antigen processing; TCR, T-cell receptor; V(D)J, variable/diversity/joining.
Adapted with permission from Bolognia JL, Jorizzo JL, Rapini RP, eds. *Dermatology*, 2nd edn. London: Elsevier, 2007.

TABLE I. Ectodermal Dysplasias of Group A (N = 163), Modified From Visinoni et al. [2009]

	Ectodermal dysplasia (ED)	Reference (OMIM, whenever available)	Inheritance
	Subgroup hair–teeth–nails–sweat glands. N = 38		
1	Alopecia-contractures-dwarfism mental retardation syndrome	203550	AR
2	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome (AEC; Hay–Wells syndrome); **Rapp–Hodgkin syndrome (129400) included**	106260	AD
3	Anonychia with flexural pigmentation	106750	AD
4	AREDYLD (Acrorenal field defect, ED, and lipoatrophic diabetes)	207780	AR
5	Arthrogryposis and ED	601701	AR
6	Camarena syndrome	Freire-Maia and Pinheiro [1984]	AD?; XD?
7	Cleft lip/palate-ED syndrome (CLPED1; Zlotogora–Ogur syndrome; Margarita Island syndrome)	225060	AR
8	Curly hair-acral keratoderma-caries syndrome	van Steensel and van der Hout [2009]	AD
9	Dyskeratosis congenita, AD (Dyskeratosis congenita, Scoggins type)	127550	AD
10	Dyskeratosis congenita, AR	224230	AR
11	Dyskeratosis congenita, X-linked (Zinsser–Cole–Engman syndrome)	305000	XR
12	Ectrodactyly, ED, and cleft lip/palate syndrome (EEC1)	129900	AD
13	Ectrodactyly, ED, and cleft lip/palate syndrome 3 (EEC3)	604292	AD
14	ED hypohidrotic, with acanthosis nigricans (Lelis syndrome)	608290	?
15	**ED-syndactyly syndrome 2 (EDSS2)**	613576	AR
16	ED with natal teeth, Turnpenny type	601345	AD
17	ED, Caratinga type	Montebelo et al. [1996]	AD?; XD?
18	ED, hypohidrotic, with hypothyroidism and agenesis of the corpus callosum	225040	AD?; AR?; XD?
19	Focal dermal hypoplasia (FDH)	305600	XD
20	Hypohidrotic ED, autosomal dominant (ADHED)	129490	AD
21	Hypohidrotic ED, autosomal recessive (ARHED)	224900	AR
22	Hypohidrotic ED, X-linked (XLHED; Christ–Siemens–Touraine syndrome; CST syndrome)	305100	XR
23	Hypohidrotic ED with immune deficiency	300291	XD
24	Hypohidrotic ED with immunodeficiency, osteopetrosis, and lymphedema (OLEDAID syndrome)	300301	XD
25	Hypomelanosis of Ito (HMI, Incontinentia pigmenti type I; IP1)	300337	XD
26	Keratitis-ichthyosis-deafness syndrome, autosomal dominant (KID syndrome, AD)	148210	AD
27	Keratitis-ichthyosis-deafness syndrome, autosomal recessive (KID syndrome, AR)	242150	AR
28	Naegeli syndrome (Naegeli–Franceschetti–Jadassohn syndrome, NFJS)	161000	AD
29	Odontoonychodermal dysplasia (OODD); **Schöpf–Schulz–Passarge syndrome (224750) included**	257980	AR
30	Odontotrichomelic syndrome (tetramelic deficiencies, ectodermal dysplasia, deformed ears, and other abnormalities)	273400	AR
31	Pachyonychia congenita, type 1 (PC1)	167200	AD
32	Pachyonychia congenita, type 2 (PC2)	167210	AD
33	Papillon–Lefevre syndrome	245000	AR
34	Rosselli–Gulienetti syndrome	225000	AR
35	Scalp-ear-nipple syndrome (Finlay–Marks syndrome; ED with adrenal cyst)	181270; 129550	AD
36	Tricho-odonto-onychodysplasia with pili torti	Freire-Maia and Pinheiro [1984]	AD?; XD?
37	Tricho-onycho-dental dysplasia (TOD)	Koshiba et al. [1978]	AD
38	Xeroderma-talipes-enamel defects (XTE)	Moynahan [1970]	AR
	Subgroup hair–teeth–nails. N = 34		
39	Ackerman syndrome	200970	AR
40	ADULT syndrome	103285	AD
41	Arthrogryposis, ED, cleft lip/palate, and developmental delay	301815	XR
42	Cardiofaciocutaneous syndrome (CFC)	115150	AD
43	**Cardiomyopathy, dilated, with woolly hair, and keratoderma (Carvajal syndrome)**	605676	AR
44	Clouston syndrome (ectodermal dysplasia, hidrotic, autosomal dominant)	129500	AD

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TABLE I. (*Continued*)

	Ectodermal dysplasia (ED)	Reference (OMIM, whenever available)	Inheritance
45	Coffin–Siris syndrome	135900	AD?; AR?; XD?
46	Costello syndrome	218040	AR
47	**Cranioectodermal dysplasia (CED1, Sensenbrenner syndrome); CED2 (613610), CED3 (614099) and CED4 (614378) included**	218330	AR
48	Dermoodontodysplasia	125640	AD
49	Dolichocephaly, dental defects, trichodysplasia	Freire-Maia and Pinheiro [1984]	AD
50	**ED-syndactyly syndrome 1 (EDSS1); ED with pillous anomaly and syndactyly (**Wiedemann et al., 1978**) included**	613573	AR
51	ED, trichoodontoonychial type	129510	AD
52	Ellis–van Creveld syndrome (EVC)	225500	AR
53	GAPO syndrome (growth retardation, alopecia, pseudoanodontia, and optic atrophy)	230740	AR
54	GOMBO syndrome	233270	AR
55	Hidrotic ED autosomal recessive (Fried's tooth and nail syndrome)	602401	AR
56	Hypotrichosis with pili bifurcate	Beemer et al. [1987f]	AR?
57	Incontinentia pigmenti (IP2)	308300	XD
58	Oculotrichodysplasia (OTD)	257960	AR
59	Odonto-onychodysplasia-alopecia	Pinheiro and Freira Maia [1981]	AR
60	Odontotrichoungual-digital-palmar syndrome	601957	AD?; XD?
61	Pineal hyperplasia, insulin-resistant diabetes mellitus, and somatic abnormalities	262190	AR
62	Rothmund–Thomson syndrome (RTS)	268400	AR
63	Schinzel–Giedion midface-retraction syndrome	269150	AR?; AD?
64	Sener syndrome	606156	?
65	**Split-hand/foot malformation (SHFM1); SHFM3 (246560), SHFM4 (605289), and SHFM5 (606708) included**	183600	AD
66	Thumb deformity and alopecia	188150	AD
67	Trichodentoosseus syndrome (TDO)	190320	AD
68	Tricho-dermodysplasia-dental defects	Pinheiro et al. [1986]	AD?; XD?
69	Trichoodontoonychial dysplasia with bone deficiency	275450	AR?
70	Trichorhinophalangeal syndrome, type I (TRPS1)	190350	AD
71	Trichothiodystrophy, photosensitive (TTDP)	601675	AR
72	Witkop syndrome	189500	AD
	Subgroup hair–teeth–sweat glands. N = 8		
73	Böök syndrome	112300	AD
74	Cleft lip/palate, ED, acral anomalies	Richieri-Costa et al. [1992]	AR
75	Hypohidrotic ED with focal sweating	Gorlin [1988]	AR?; XR?
76	IFAP syndrome with or without bresheck syndrome (Ichthyosis follicularis, atrichia, and photophobia syndrome)	308205	XR
77	Johnson neuroectodermal syndrome	147770	AD
78	Lenz–Passarge dysplasia	Lenz [1963]	XD
79	Leukomelanoderma, infantilism, mental retardation, hypodontia, hypotrichosis	246500	AR
80	Ulnar-mammary syndrome (UMS)	181450	AD
	Subgroup hair–nails–sweat glands. N = 4		
81	Alopecia-skin atrophy-anonychia-tongue defects	Sequeiros and Sack [1985]	?
82	ED, hypohidrotic, with hypothyroidism and ciliary dyskinesia (HEDH syndrome)	225050	AR
83	ED/skin fragility syndrome	604536	AR
84	Fischer–Volavsek syndrome	Fischer [1921]	AD
	Subgroup teeth–nails–sweat glands. N = 2		
85	Ameloonychohypohidrotic syndrome	104570	AD
86	Limb-mammary syndrome (LMS)	603543	AD
	Subgroup hair–teeth. N = 29		
87	Barber–Say syndrome	209885	AR?; AD?; XD?
88	Blepharocheilodontic syndrome	119580	AD
89	Brachymetapody-anodontia-hypotrichosis-albinoidism	211370	AR
90	CAHMR syndrome (Cataract, hypertrichosis, mental retardation syndrome)	211770	AR
91	Cerebellar ataxia and ED	212835	AR

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TABLE I. (*Continued*)

	Ectodermal dysplasia (ED)	Reference (OMIM, whenever available)	Inheritance
92	Cleft lip/palate-oligodontia-syndactyly-hair defects	Martínez et al. [1987]	AD?; XD?
93	Coloboma, congenital heart disease, ichthyosiform dermatosis, mental retardation, and ear anomalies syndrome (CHIME, Zunich neuroectodermal syndrome)	280000	AR
94	Congenital atrichia, palmoplantar hyperkeratosis, mental retardation, and early loss of teeth	Steijlen et al. [1994]	AR?
95	Dubowitz syndrome	223370	AR
96	ED and neurosensory deafness	224800	AR
97	ED, Cape Verde	Werninghaus [1993]	AR
98	EEM syndrome (ED, ectrodactyly, and macular dystrophy); **Hypotrichosis, congenital, with juvenile macular dystrophy (HJMD, 601553) included**	225280	AR
99	Gorlin–Chaudhry–Moss syndrome	233500	AR
100	Hallermann–Streiff syndrome (HSS)	234100	AR
101	Hypertrichosis terminalis, generalized, with our without gingival hyperplasia (Gingival fibromatosis with hypertrichosis)	135400	AD
102	Hypertrichosis universalis	145700	AD
103	Johanson–Blizzard syndrome (JBS)	243800	AR
104	Mental retardation, hypotrichosis, and syndactyly	Lopes and Marques-de-Faria [1996]	AR?
105	Oculodentoosseous dysplasia, recessive	257850	AR
106	Oculodentodigital dysplasia (ODDD)	164200	AD
107	Orofaciodigital syndrome I (OFD1)	311200	XD
108	Pili torti, early onset	261900	AR
109	Pilodental dysplasia with refractive errors	262020	AR
110	Progeroid short stature with pigmented nevi (Mulvihill–Smith syndrome)	176690	AD
111	Rodrigues blindness (Microphthalmia, microcornea, and sclerocornea with short stature and hair and dental abnormalities)	268320	AR
112	Trichodental dysplasia	601453	AD
113	Trichodysplasia and amelogenesis imperfect	Angelos and Jorgenson [1993]	AD?; XD?
114	Uncombable hair, retinal pigmentary dystrophy, dental anomalies, and brachydactyly	191482	AD
115	Walbaum–Dehaene–Schlemmer syndrome	Walbaum et al. [1971]	AR
	Subgroup hair–nails. N = 25		
116	Anonychia-onychodystrophy with hypoplasia or absence of distal phalanges (Cooks syndrome)	106995	AD
117	Autosomal recessive neurodegenerative disorder with trichorrhexis invaginata and ED	Gyure et al. [1992]	AR?
118	Cartilage-hair hypoplasia (CHH)	250250	AR
119	Curly hair-ankyloblepharon-nail dysplasia syndrome (CHANDS)	214350	AR
120	ED hidrotic, Christianson–Fourie type	601375	AD
121	ED with skin anomalies and mental retardation	Halal et al. [1991]	AR
122	ED, "pure" hair-nail type	602032	AD?
123	Hair-nail dysplasia	Pinheiro and Freire-Maia [1992]	AD
124	Hairy elbows (hypertrichosis cubiti)	139600	AD
125	Ichthyosis and male hypogonadism	308200	XR?
126	Ichthyosis with alopecia, eclabion, ectropion, and mental retardation	242510	AR
127	Lymphedema-hypoparathyroidism syndrome	247410	AR?; XR?
128	Monilethrix	158000	AD
129	Onychotrichodysplasia and neutropenia	258360	AR
130	Palmoplantar keratoderma and congenital alopecia, autosomal dominant (alopecia congenita with keratosis palmoplantaris)	104100	AD
131	Pili torti and onychodysplasia	Beare [1952]	AD
132	Pili torti, alopecia and onychodysplasia	Calzavara-Pinton et al. [1991]	AR
133	Polyposis, skin pigmentation, alopecia, and fingernail changes	175500	?
134	Popliteal pterygium syndrome, lethal type	263650	AR
135	**Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis syndrome (SOFT syndrome)**	Sarig et al. [2012]	AR
136	Syndrome of accelerated skeletal maturation, failure to thrive and peculiar face (Marshall syndrome II)	Marshall et al. [1971]	AR?; XR?

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TABLE I. (*Continued*)

	Ectodermal dysplasia (ED)	Reference (OMIM, whenever available)	Inheritance
137	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	601705	AR?
138	Trichomegaly with mental retardation, dwarfism, and pigmentary degeneration of retina	275400	AR
139	Tricho-onychodysplasia-xeroderma	Freire-Maia et al. [1985]	AR
140	Trichothiodystrophy, nonphotosensitive 1 (TTDN1)	234050	AR
	Subgroup hair–sweat glands. N = 4		
141	Dry skin and extranumerary areolae	Freire-Maia and Chautard-Freire-Maia [1990]	AD
142	Focal facial dermal dysplasia (Brauer syndrome); Facial ED (Setleis syndrome, 227260) included	136500	AD
143	Short stature-kidney insufficiency-ophthalmological anomaly-growth retardation-ED (SKORED)	Greenstein et al. [1985]	AR?; XR?
144	Tricho-facio-hypohidrotic syndrome	Antley et al. [1976]	AR?; XR?
	Subgroup teeth–nails. N = 14		
145	Corneodermatoosseous syndrome (CDO syndrome)	122440	AD
146	Deafness, congenital, and onychodystrophy, autosomal dominant	124480	AD
147	Deafness, onychodystrophy, osteodystrophy, and mental retardation syndrome (DOOR syndrome)	220500	AR?; AD?
148	Dermatoosteolysis, Kirghizian type	221810	AR
149	Haim–Munk syndrome (HMS)	245010	AR
150	Hearing loss, sensorineural, with enamel hypoplasia and nail defects (Heimler syndrome)	234580	AR
151	**Khan et al. chondroectodermal dysplasia**	Khan et al. [2012]	AR
152	Lacrimoauriculodentodigital syndrome (LADD)	149730	AD
153	Odontomicronychial dysplasia	601319	AR
154	Odonto-ungueal dysplasia	Pinheiro and Freire-Maia [1996]	AD
155	Otopalatodigital syndrome, type I (OPD1)	311300	XD
156	Pycnodysostosis	265800	AR
157	Weyers acrofacial dysostosis	193530	AD
158	Williams–Beuren syndrome (WBS)	194050	AD
	Subgroup teeth–sweat glands. N = 3		
159	Hypohidrotic ED with mydriasis, iris atrophy, and mental retardation	Beyer et al. [1979]	AD?
160	Kohlschutter–Tonz syndrome (epilepsy, dementia, and amelogenesis imperfecta)	226750	AR?; XR?
161	Marshall syndrome I	154780	AD
	Subgroup nail–sweat glands. N = 2		
162	Adermatoglyphia with congenital facial milia and acral blisters, digital contractures, and nail abnormalities (ED, absent dermatoglyphic pattern, changes in nails, and simian crease)	129200	AD
163	Pachyonychia congenita, autosomal recessive	260130	AR

Note: the bold text highlights information not listed in Visinoni et al. [2009].
AR, autosomal recessive; AD, autosomal dominant; XD, X-linked dominant; XR, X-linked recessive; ?, unknown.

we have consulted the OMIM, PUBMED, and Science Direct online databases.

RESULTS AND DISCUSSION

After the first International Conference on Ectodermal Dysplasias Classification, held in Charleston (South Carolina) in 2008, we listed 186 disorders in an article published in a special issue of the *American Journal of Medical Genetics* [Visinoni et al., 2009]. Within this reported series of ectodermal dysplasias, descriptions of 26 single cases were included because of their significant frequency of conditions (13.8%), and also because that by mentioning these cases the description of similar patients could be encouraged, which could possibly help clarify some poorly understood clinical presentations.

Reviewing the data and following what was decided at the 2012 International Conference on Ectodermal Dysplasias Classification (Charleston, SC), we removed single cases from the list of ectodermal dysplasias.

Table I provides a list of 163 dysplasias belonging to Group A. Besides the exclusion of the single cases and the inclusion of new EDs, new lumpings of conditions were added to this table, and the syndrome nomenclature was reviewed. For 78.5% of these conditions there is an OMIM entry, providing easy access to the more relevant information. The pattern of inheritance is also shown in Table I. It is remarkable that 39.9% are autosomal recessive, 33.7% are autosomal dominant, and 7.4% are X linked disorders. It is also noteworthy that the pattern of inheritance is not established in 19% of the ectodermal dysplasias.

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TABLE II. Ectodermal Dysplasias (N = 7) Not Included in Visinoni et al. [2009]

ED	Reference	Inheritance	Hair	Teeth	Nails	Sweat glands	Other signs
Subgroup hair–teeth–nails–sweat glands							
Cardiomyopathy, dilated, with woolly hair and keratoderma (Carvajal syndrome)	605676	AR	Woolly, wavy, wiry and auburn hair; sparse eyebrows and eyelashes	Hypodontia; diminutive upper lateral incisors; prepubertal periodontitis; premature root resorption of primary teeth	Small and thickened	Hypohidrosis	Dilated cardiomyopathy leading to congestive heart failure; epidermolytic palmoplantar keratoderma
Subgroup hair–teeth–nails							
Cranioectodermal dysplasia (CED1, Sensenbrenner syndrome); CED2 (613610), CED3 (614099) and CED4 (614378) included	218330	AR	Sparse; fine and slow-growing hair	Hypodontia; microdontia; abnormally shaped	Short and broad	Normal	Craniosynostosis; microcephaly; hypoplasic posterior corpus callosum; postnatal growth retardation; narrow thorax with pectus excavatum; short limbs; brachydactyly; nephropathy; photophobia
ED-syndactyly syndrome 1 (EDSS1); ED with pillous anomaly and syndactyly [Wiedemann et al., 1978] included	613573	AR	Sparse, coarse and breakable scalp and body hair, eyebrows and eyelashes; progressive hair loss; alopecia; pilli torti	Enamel hypoplasia; peg-shaped; widely spaced	Hypoplastic	Normal	Cutaneous syndactyly; palmar hyperkeratosis
ED-syndactyly syndrome 2 (EDSS2)	613576	AR	Sparse and slow growing scalp hair; sparse eyebrows and eyelashes, sparse to absent armpit and pubic hair; thin to absent body hair; follicular hyperkeratosis	Enamel hypoplasia; conical; widely spaced	Hypoplastic; thickened, flat, and yellowish or discolored	Hyperhidrosis (hands, face, and scalp)	Cutaneous syndactyly; palmoplantar keratoderma; hard scaly skin; pointed nose, thin upper lips, and large prominent ear pinnae; cardiomegaly
Split-hand/foot malformation (SHFM1); SHFM3 (246560), SHFM4 (605289) and SHFM5 (606708) included	183600	AD	Sparse; alopecia	Hypodontia	Dysplasic	Normal	Mental retardation; ectrodactyly; cleft lip/palate; micrognathia; occasional deafness and cardiac defects
Subgroup hair–nails							
Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis syndrome (SOFT syndrome)	Sarig et al. [2012]	AR	Hypotrichosis; postpubertal sparse and short hair	Normal	Hypoplastic	Normal	Short stature; short and thick long bones; hypoplastic pelvis and sacrum; clinodactyly and brachydactyly; hypoplastic distal phalanges; peculiar faces; small ears; high-pitched voice
Subgroup teeth–nails							
Khan et al. chondroectodermal dysplasia	Khan et al. [2012]	AR	Normal	Absence of mandibular central incisors; misshaped; proeminent	Dysplastic; enlarged nail bed; hypertrophic and convex shaped	Normal	Short stature; osteopenia;with breakable bonés; night blindness; watering eyes; ear infections

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TABLE III. Genes (N = 13[a]) and Chromosomal Regions (N = 6) Responsible for 15 Different Ectodermal Dysplasias Not Included in Visinoni et al. [2009]

Chromossome	Gene	Protein or gene product	Ectodermal dysplasia (ED)	OMIM/Refs
Xp22.12–p22.11	*MBTPS2*	Membrane-bound transcription factor site-2 protease	IFAP syndrome with or without bresheck syndrome	308205
1q23.3	*PVRL4*	Poliovirus receptor-related protein 4	ED-syndactyly syndrome 1 (EDSS1)	613573
2p24.1	*WDR35*	WD repeat-containing protein 35	Cranioectodermal dysplasia (CED1–4)	218330, 613610, 614099, 614378
2q24.1–q31.1			Khan et al. chondroectodermal dysplasia	Khan et al. [2012]
2q31			SHFM1, 3–5 syndrome	183600, 246560, 605289, 606708
2q37.3	*TWIST2*	Twist-related protein 2	Focal facial dermal dysplasia (facial ED)	136500, 227260
3p21.2	*POC1A*	Centriolar protein homolog A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis syndrome (SOFT)	Sarig et al. [2012]
3q21.3–q22.1	*IFT122*	Intraflagellar transport protein 122 homolog	Cranioectodermal dysplasia (CED1–4)	218330, 613610, 614099, 614378
4p14	*WDR19*	WD repeat-containing protein 19	Cranioectodermal dysplasia (CED1–4)	218330, 613610, 614099, 614378
6p24.3	*DSP*	Desmoplakin	Cardiomyopathy, dilated, with woolly hair and keratoderma (Carvajal syndrome)	605676
6q22.31	*GJA1*[a]	Connexin 43	Oculodentodigital dysplasia (ODDD)	164200
			Hallermann–Streiff syndrome (HSS)	234100
7p21.2–p14.3			ED syndactyly syndrome 2 (EDSS2)	613576
7q21.2–q21.3			SHFM1, 3–5 syndrome	183600, 246560, 605289, 606708
10q24			SHFM1, 3–5 syndrome	183600, 246560, 605289, 606708
14q24.3	*IFT43*	Intraflagellar transport protein 43 homolog	Cranioectodermal dysplasia (CED1–4)	218330, 613610, 614099, 614378
16p13.3	*ROGDI*	Protein rogdi homolog	Kohlschutter–Tonz syndrome (KTZS)	226750
17p11.2	*PIGL*	N-acetylglucosaminyl-phosphatidylinositol de-N-acetylase	Coloboma, congenital heart disease, ichthyosiform dermatosis, mental retardation, and ear anomalies syndrome (CHIME)	280000
17q24.2–q24.3			Hypertrichosis terminalis, generalized, with our without gingival hyperplasia	135400
18q12.3	*SETBP1*	SET-binding protein	Schinzel–Giedion midface retraction synsdrome	269150
21q22.3	*RIPK4*	Receptor-interacting serine-threonine-protein kinase 4	Popliteal pterygium syndrome, lethal type	263650

[a]The *GJA1* gene was related only to Oculodentodigital dysplasia (ODDD) in Visinoni et al. [2009].

With the description of new syndromes and the better delineation of the conditions known, some syndromes initially described as different entities were considered variations of the same disorder. The opposite also occurs: descriptions considered as being the same clinical entity may be separated in two or more different conditions. This lumping and splitting of syndromes as well as the reports of previously undescribed conditions make the classifications dynamic and modifiable by incorporating the new knowledge. For instance, the TP63-related syndromes, ankyloblepharon-ectodermal defects-cleft lip and palate (AEC or Hay–Wells syndrome, OMIM 106260) and the Rapp–Hodgkin syndrome (OMIM 129400) are now considered as being different phenotypic expressions of the same condition [Prontera et al., 2008; Clements et al., 2010]. Furthermore, Prontera et al. [2011] suggested that ADULT

15524833, 2014, 10, Downloaded from https://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.36616 by Egyptian National Sci. Network (Enstinet), Wiley Online Library on [12/12/2025]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

EPIDERMOLYSIS BULLOSA: SIMPLEX, JUNCTIONAL AND DYSTROPHIC SUBTYPES AND KINDLER SYNDROME		
Subtype	**Inheritance**	**Defective protein(s)**
Suprabasal intraepidermal cleavage		
Acral peeling skin syndrome	AR	Transglutaminase 5
EB superficialis	Unknown	Unknown
Acantholytic EBS	AR	Desmoplakin; plakoglobin
Skin fragility syndromes		
Desmoplakin deficiency (skin fragility–woolly hair syndrome)	AR	Desmoplakin
Plakoglobin deficiency	AR	Plakoglobin
Plakophilin deficiency (skin fragility–ectodermal dysplasia syndrome)	AR	Plakophilin 1
Basal intraepidermal cleavage		
EBS, localized (formerly Weber–Cockayne)	AD	Keratins 5 and 14
EBS, generalized severe (formerly Dowling–Meara)		
EBS, generalized intermediate		
EBS with mottled pigmentation	AD	Keratin 5
EBS, migratory circinate		
EBS, AR-KRT14	AR	Keratin 14
EBS due to KLHL24	AD	Kelch-like protein 24
EBS with muscular dystrophy	AR	Plectin
EBS with pyloric atresia	AR	Plectin; $\alpha_6\beta_4$ integrin
EBS-Ogna	AD	Plectin
EBS, AR-BP230 deficiency	AR	Bullous pemphigoid antigen 1 (BP230)
EBS, AR-exophilin 5 deficiency	AR	Exophilin 5
Intra-lamina lucida cleavage		
JEB, generalized severe (formerly Herlitz)	AR	Laminin 332
JEB, generalized intermediate	AR*	Laminin 332; collagen XVII
JEB with pyloric atresia	AR	$\alpha_6\beta_4$ integrin
JEB, late onset	AR	Collagen XVII
JEB with respiratory and renal involvement	AR	α_3 integrin subunit
JEB, localized	AR	Collagen XVII; $\alpha_6\beta_4$ integrin
JEB, inversa	AR	Laminin 332
JEB-laryngo-onycho-cutaneous syndrome	AR	Laminin α3 chain (a isoform)
Sublamina densa cleavage		
DDEB, generalized	AD	Collagen VII
Other DDEB variants: acral, pretibial, pruriginosa, nails only		
DDEB, bullous dermolysis of the newborn		
RDEB, generalized severe (formerly Hallopeau–Siemens)	AR	
RDEB, generalized intermediate		
RDEB, inversa		
Other RDEB variants: localized, pretibial, pruriginosa, centripetalis		
RDEB, bullous dermolysis of the newborn		
Mixed cleavage planes		
Kindler syndrome	AR	Kindlin-1
*One case with AD inheritance has been reported.		

Table 32.1 Epidermolysis bullosa (EB): simplex, junctional and dystrophic subtypes and Kindler syndrome. Entities with a lighter background represent rare variants. AD, autosomal dominant; AR, autosomal recessive; EBS, epidermolysis bullosa simplex; DDEB, dominant dystrophic EB; RDEB, recessive dystrophic EB.

key component of the lamina lucida of the dermal–epidermal junction (see Ch. 28)[16]. In laryngo-onycho-cutaneous syndrome, the underlying mutations affect only the a isoform of the laminin α3 subunit. The milder generalized intermediate form of JEB (JEB-gen/intermed, previously known as JEB-non-Herlitz and generalized atrophic benign EB) results from mutations within the genes for either a subunit of laminin 332 or type XVII collagen. JEB with pyloric atresia, which is more common than EBS with pyloric atresia, is also caused by mutations within either of the two genes that encode the subunits of $\alpha_6\beta_4$ integrin. A newly recognized form of JEB associated with respiratory and renal involvement results from mutations in the integrin α_3 chain.

Dystrophic EB

Dystrophic EB (DEB) is transmitted in either an autosomal dominant or autosomal recessive manner and is caused by mutations in the type VII collagen gene. Dominant DEB (DDEB) results from dominant-negative mutations, typically a missense mutation that leads to substitution of

Table 6.2 Revised classification of inherited epidermolysis bullosa (EB)

Major EB type	Subtype	Gene defect	Target protein
Simplex (EBS)			
– suprabasal	Lethal acantholytic	*DSP*	Desmoplakin
	Plakophilin deficiency	*PKP1*	Plakophilin-1
	Superficialis	*?*	?
– basal	Localized (Weber–Cockayne)	*KRT5, 14*	K5, 14
	Dowling–Meara	*KRT5, 14*	K5, 14
	Other generalized (includes Koebner)	*KRT5, 14*	K5, 14
	EBS, with mottled pigmentation	*KRT5*	K5
	EBS, with muscular dystrophy	*PLEC1*	Plectin
	EBS, with pyloric atresia	*PLEC1, ITGA6, ITGB4*	Plectin, α6β4 integrin
	Autosomal recessive	*KRT14*	K14
	Ogna type	*PLEC1*	Plectin
	Migratory circinate	*KRT5*	K5
Junctional (JEB)			
– Herlitz	Herlitz	*LAMA3, LAMB3, LAMC2*	Laminin 5
– others	JEB, non Herlitz – generalized	*LAMA3, LAMB3, LAMC2,*	Laminin 5, type XVII collagen
	JEB, non Herlitz – localized	*COL17A1*	Type XVII collagen
	JEB, with pyloric atresia	*COL17A1*	α6β4 integrin
	JEB, inversa	*ITG176, ITGB4*	Laminin 5
	JEB, late onset	*LAMA3, LAMB3, LAMC2(?)*	?
	LOC syndrome	*?*	Laminin 332 (α3 chain)
		LAMA3	
Dystrophic (DEB)			
– dominant (DDEB)	DDEB, generalized	*COL7A1*	Type VII collagen
	DDEB, acral	*COL7A1*	Type VII collagen
	DDEB, pretibial	*COL7A1*	Type VII collagen
	DDEB, pruriginosa	*COL7A1*	Type VII collagen
	DDEB, nails only	*COL7A1*	Type VII collagen
	DDEB, bullous dermolysis of newborn	*COL7A1*	Type VII collagen
– recessive (RDEB)	RDEB, severe generalized	*COL7A1*	Type VII collagen
	RDEB, generalized other	*COL7A1*	Type VII collagen
	RDEB, inversa	*COL7A1*	Type VII collagen
	RDEB, pretibial	*COL7A1*	Type VII collagen
	RDEB, pruriginosa	*COL7A1*	Type VII collagen
	RDEB, centripetalis	*COL7A1*	Type VII collagen
	RDEB, bullous dermolysis of newborn	*COL7A1*	Type VII collagen
Kindler's syndrome	Kindler's syndrome	*KIND1*	Kindlin 1

ratories in any country will maintain stocks of these antibodies. The molecular defects, as currently known,[858] are listed in **Table 6.2**. A proposal to add a subgroup of hemidesmosomal cases, to include the plectin-deficient variant of epidermolysis bullosa simplex with muscular dystrophy, has not been incorporated into the most recent classification.[852,859,860] The intraepidermal/epidermolytic (simplex) group, which involves lysis of basal cells, will be considered first. Before doing so, brief mention will be made of the atypical nevi that may develop in all subsets of epidermolysis bullosa, particularly the recessively inherited variants.[861] The dermoscopic pattern may mimic melanoma but the absence of features such as irregular streaks, a blue-whitish veil, regression features, or black dots gives a distinctive dermoscopic pattern.[861]

Epidermolysis bullosa simplex

Epidermolysis bullosa simplex (EBS), one of the four major subgroups of epidermolysis bullosa (if Kindler's syndrome is accepted as the fourth category), is a mechanobullous disorder characterized by intraepidermal cleavage, usually through the basal layer of cells.[839,862] There are several distinct clinical variants.[863] Most cases have a mild clinical expression with blisters which heal without scarring, and an autosomal dominant inheritance.[864,865] Rare cases with an autosomal recessive inheritance and a more severe clinical course, sometimes with associated neuromuscular disease (particularly muscular dystrophy), have been reported.[866–868] Not all recessive forms have severe disease.[869,870] A prenatal diagnosis was made in the past using fetal skin biopsy samples.[822,871,872] Analysis of fetal DNA using chorionic villi or amniotic fluid cells is now the preferred technique.[873–876] Keratinocyte cell lines have been established for EBS, providing a potential source for in-vitro experiments using gene therapy.[877]

The molecular basis of most of the simplex subgroup resides in defects that disrupt the assembly, structure and/or function of the keratin intermediate filament skeleton of the basal keratinocyte – the coexpressed peptides keratin 5 and 14.[878–884] The keratin 5 (*KRT5*) gene maps to 12q13 and the keratin 14 (*KRT14*) gene to 17q12–q21. It appears that mutations in certain locations of the keratin genes (usually the ends of conserved α-helical rod domains) are more likely to produce the severe forms, such as the Dowling–Meara type (see below).[885] Twelve subtypes of EBS are recognized in the latest classification (see Table 6.2), which is based on the Report of the Third International Consensus Meeting held in 2007.[852] Only the four major subtypes of EBS will be considered in detail.

Other generalized type (includes Koebner variant)

The generalized (Koebner – OMIM 131900) clinical variant (now included in the 'other generalized type') is characterized by the devel-

Epidermolysis Bullosa

Table 23 Classification of epidermolysis bullosa

Major EB type	Major EB subtypes	Targeted protein(s)
EB simplex (EBS)	Suprabasal subtypes	
	• Lethal acantholytic EBS	Desmoplakin
	• Plakophilin-1 deficiency	Plakophilin-1
	• EBS superficialis (EBSS)	unknown
	Basal subtypes	
	• EBS, localized (Weber–Cockayne)	K5, K14
	• EBS, Dowling–Meara	K5, K14 – clumped tonofilaments on EM
	• EBS, other generalized (Koebner)	K5, K14
	• EBS with mottled pigmentation	K5
	• EBS with muscular dystrophy	Plectin
	• EBS with pyloric atresia (MC with JEB)	Plectin, α6β4 integrin
	• EBS, autosomal recessive	K14
	• EBS, Ogna variant	Plectin
	• EBS, migratory circinate	K5
Junctional EB(JEB)	JEB, Herlitz type	Laminin-332
	JEB, generalized non-Herlitz	Laminin-332; type XVII collagen
	JEB, localized non-Herlitz	Type XVII collagen
	JEB with pyloric atresia	α6β4 integrin
	JEB, inversa	Laminin-332
	JEB, late onset	unknown
	LOC (*l*aryngo-*o*nycho-*c*utaneous) syndrome (AKA -Shabbir's syndrome)	Laminin-332 (α3 chain)
Dominant dystrophic EB (DDEB)	DDEB, generalized	Type VII collagen
	DDEB, acral	
	DDEB, pretibial	
	DDEB, pruriginosa	
	DDEB, nails only	
	DDEB, bullous dermolysis of newborn (not always transient)	
Recessive dystrophic EB (RDEB)	RDEB, severe generalized (Hallopeau–Siemens)	Type VII collagen
	RDEB, generalized other (non-HS)	
	RDEB, inversa	
	RDEB, pretibial	
	RDEB, pruriginosa	
	RDEB, centripetalis	
	RDEB, bullous dermolysis of newborn (not always transient)	
Kindler syndrome		Kindlin-1

Table 8.1 List of the ichthyoses and related disorders

TYPES	SYNONYM
Non-syndromic ichthyoses	
Autosomal recessive congenital ichthyosis	ARCI
Lamellar ichthyosis	LI; ichthyosis congenita type 2
Congenital ichthyosiform erythroderma	CIE; nonbullous congenital ichthyosiform erythroderma; NBCIE, ichthyosis congenita type 1
Harlequin ichthyosis	HI; harlequin baby, harlequin fetus
Intermediate LI/CIE phenotypes	Non-classified ARCI
Self-healing collodion baby	SHCB
Bathing suit ichthyosis	BSI
Autosomal dominant congenital ichthyosis	
Autosomal dominant lamellar ichthyosis/ Autosomal dominant congenital ichthyosiform erythroderma	adLI; adCIE
Ichthyosis en confettis	Ichthyosis variegata, congenital reticular ichthyosiform erythroderma
Ichthyosis vulgaris	IV
Keratinopathic ichthyosis	KPI
Epidermolytic ichthyosis	EI; epidermolytic hyperkeratosis; EHK; bullous congenital ichthyosiform erythroderma of Brocq; bullous ichthyosis
Superficial epidermolytic ichthyosis	SEI; ichthyosis bullosa of Siemens; exfoliative ichthyosis
Mosaic epidermolytic ichthyosis	Mosaic EI, mosaic EHK; epidermal nevus of epidermolytic type
Annular epidermolytic ichthyosis	AEI
Ichthyosis Curth–Macklin	ICM; ichthyosis hystrix Curth–Macklin
KLICK syndrome	Keratosis Linearis with Ichthyosis Congenita and sclerosing Keratoderma
Loricrin keratoderma	LK; mutilating keratoderma with ichthyosis
Peeling skin syndrome	Keratolysis exfoliativa congenita; continual skin peeling; peeling skin syndrome type A/B, peeling skin disease, PSD
X-linked recessive ichthyosis	RXLI, steroid sulfatase deficiency
Syndromic ichthyoses	
ARC syndrome	Arthrogryposis, renal dysfunction, and cholestasis syndrome
CEDNIK syndrome	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma
Chondrodysplasia punctata, X-linked dominant	Conradi–Hünermann–Happle syndrome
Ichthyosis hypotrichosis syndrome	HIS; congenital ichthyosis, follicular atrophoderma, hypotrichosis, and hypohidrosis
Ichthyosis–hypotrichosis–sclerosing cholangitis syndrome	IHSC syndrome; neonatal ichthyosis-sclerosing cholangitis syndrome; NISCH syndrome; ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis
Ichthyosis prematurity syndrome	IPS
IFAP syndrome	Ichthyosis Follicularis, Atrichia, and Photophobia syndrome
Infantile Gaucher disease	Gaucher disease type II; GD II; Gaucher disease of the acute neuronopathic type
KID syndrome (including HID syndrome)	Keratitis–Ichthyosis-like–Deafness syndrome Ichthyosiform erythroderma, corneal involvement, and deafness, autosomal dominant (hystrix-like ichthyosis-deafness syndrome; ichthyosis hystrix gravior, type Rheydt)
MEDNIK syndrome	Mental retardation, Enteropathy, Deafness, peripheral Neuropathy, Ichthyosis, and Keratodermia syndrome
Multiple sulfatase deficiency	MSD
Netherton syndrome/ichthyosis linearis circumflexa	NS; NTS; Comèl–Netherton syndrome
Neutral lipid storage disease with ichthyosis	Chanarin–Dorfman syndrome; triglyceride storage disease with impaired long-chain fatty acid oxidation; ichthyosiform erythroderma with leukocyte vacuolization
Refsum disease	Adult Refsum disease; heredopathia atactica polyneuritiformis; phytanic acid oxidase deficiency; hereditary motor and sensory neuropathy IV; HMSN4
Rhizomelic chondrodysplasia punctata (RCDP1)	Rhizomelic chondrodyslplasia type 1; chondrodystrophia calcificans punctata
Sjögren–Larsson syndrome	SLS
Trichothiodystrophy	Tay syndrome; (P)IBIDS syndrome: (Photosensitivity)–Ichthyosis-TTD; Brittle hair–Impaired intelligence–Decreased fertility–Short stature syndrome
Related disorders	
CHILD syndrome	Unilateral CIE
CHIME syndrome	Ocular Colobomas, congenital Heart disease, early-onset Ichthyosiform dermatosis, Mental retardation and Ear anomalies (conductive hearing loss)/Epilepsy; Zunich–Kaye syndrome; Zunich neuroectodermal syndrome
Desmons syndrome	Ichthyosiform erythroderma, corneal involvement, and deafness; autosomal recessive
Erythrokeratodermia variabilis	EKV, erythrokeratodermia figurate variabilis Mendes da Costa
Ichthyosis, hepatosplenomegaly, and cerebellar degeneration	
Ichthyosis–mental retardation syndrome with large keratohyalin granules	
Keratosis spinulosa delcalvans	KFSD
Neu-Laxora syndrome	NLS
Acquired ichthyosis	

Table 1 Nonsyndromic ichthyoses

Disease	Mode of inheritance	Gene(s)
Common ichthyoses		
- Ichthyosis vulgaris [146700]	Autosomal semidominant	*FLG*
- X-linked ichthyosis – nonsyndromic [308100]	X-linked	*STS*
Autosomal-recessive congenital ichthyoses		
- Harlequin ichthyosis		
o ARCI4B [242500]	AR	*ABCA12*
- Lamellar ichthyosis and/or **- Congenital ichthyosiform erythrodermia and/or** **- Self-improving congenital ichthyosis**		
o ARCI1 [242300] o ARCI2 [242100] o ARCI3 [606545] o ARCI4A [601277] o ARCI5 [604777] o ARCI6 [612281] o ARCI8 [613943] (late onset) o ARCI9 [615023] o ARCI10 [615024] o ARCI11 [613943] (see also Table 2) o ARCI12 [605848] o ARCI13 [609769] o ARCI14 [604125]	AR	*TGM1* *ALOX12B* *ALOXE3* *ABCA12* *CYP4F22* *NIPAL4* *LIPN* *CERS3* *PNPLA1* *ST14* *CASP14* *SDR9C7* *SULT2B1*
- Self-healing collodion baby		
o ARCI1 [242300]	AR	*TGM1*
- Bathing suit ichthyosis		
o ARCI1 [242300]	AR	*TGM1*
	AD	*ASPVR1*
Keratinopathic ichthyosis		
o Epidermolytic ichthyosis [113800]	AD	*KRT1/KRT10*
o Superficial epidermolytic ichthyosis [146800]	AD	*KRT2*
- KPI variants		
o Annular epidermolytic ichthyosis [607602]	AD	*KRT1/KRT10*
o Ichthyosis Curth–Macklin [146590]	AD	*KRT1*
o Autosomal-recessive epidermolytic ichthyosis [113800]	AR	*KRT10*
o Congenital reticular ichthyosiform erythrodermia/ichthyosis with confetti [609165]	AD	*KRT10/KRT1*
o Epidermolytic nevi [113800][a]	Somatic mutations	*KRT1/KRT10*
Peeling skin syndromes – generalized		
- Peeling skin disease type B [270300]	AR	*CDSN*
- Peeling skin disease type A [610190] [616284]		*CHST8/FLG2*
- Exfoliative ichthyosis [607936] - Exfoliative ichthyosis [617115]		*CSTA* *SERPINB8*
Other nonsyndromic ichthyoses		
- Autosomal dominant lamellar ichthyosis [146750]	AD	ASPVR1
- Keratosis linearis–ichthyosis congenita keratoderma [601952]	AR	*POMP*
- Loricrin keratoderma [604117]	AD	*LOR*
- Erythrokeratodermia variabilis [133200]	AD	*GJB3/GJB4*

AD autosomal dominant, *AR* autosomal recessive
[a]May indicate a gonadal mosaicism, which can cause generalized epidermolytic ichthyosis in the offspring generation. OMIM in [ ], http://www.ncbi.nlm.nih.gov/omim

their environment. The squamous epithelium consists of keratinocytes, whose degree of differentiation is distinguished functionally and morphologically by several histological layers (Figs. 1 and 2).

3.1 Stratum Basale (Basal Cell Layer)

Here the cells are aligned vertically. Within the layer and immediately above it (stratum suprabasale) mitosis takes

Table 2 Syndromic ichthyoses

Disease	Mode of inheritance	Gene(s)
X-linked ichthyosis syndromes		
- X-linked ichthyosis, syndromic [308100, 300500, 300533]	X-linked	*STS* (and others)
- Ichthyosis follicularis alopecia photophobia syndrome [308205]		*MBTPS2*
- Conradi–Hünermann–Happle syndrome (CDPX2) [302960]		*EBP*
Autosomal ichthyosis syndromes with prominent hair abnormalities		
- Netherton syndrome [256500]	AR	*SPINK5*
- Ichthyosis hypotrichosis syndrome [602400]		*ST14*
- Neonatal ichthyosis-sclerosing cholangitis/ichthyosis hypotrichosis sclerosing cholangitis syndrome [607626]		*CLDN1*
Autosomal ichthyosis syndromes with prominent neurological signs		
- Keratitis ichthyosis deafness syndrome [148210]	AD	*GJB2* (*GJB6*)
- Refsum syndrome (HMSN4) [266500]	AR	*PHYH/PEX7*
- Multiple sulfatase deficiency [272200]		*SUMF1*
- Gaucher syndrome type 2 [230900]		*GBA*
- Sjögren–Larsson syndrome [270200]		*ALDH3A2*
- Trichothiodystrophy/Tay syndrome o TTD1 [601675]	AR	*ERCC2/XPD*
o TTD2 [616390]		*ERCC3/XPB*
o TTD3 [616395]		*GTF2H5/TFIIH*
- Cerebral dysgenesis–neuropathy–ichthyosis–palmoplantar keratoderma syndrome [609528]	AR	*SNAP29*
- Mental retardation–enteropathy–deafness–neuropathy–ichthyosis–keratoderma syndrome [609313]		*AP1S1*
- Arthrogryposis–renal dysfunction–cholestasis syndrome [208085]		*VPS33B*
- Ichthyosis–spastic quadriplegia–mental retardation syndrome [614457]		*ELOVL4*
- Coloboma–congenital heart disease–ichthyosiform dermatosis–mental retardation–ear anomalies syndrome [280000]		*PIGL*
Autosomal ichthyosis syndromes with other prominent findings		
- Desmons syndrome [600157]		*AP1B1*
- Ichthyosis prematurity syndrome [608649]	AR	*SLC27A4* (*FATP4*)
- Severe dermatitis–multiple allergies–metabolic wasting syndrome [615508]		*DSG1* (see also *DSP*)
- Chanarin–Dorfman syndrome/neutral lipid storage disease with ichthyosis [275630]		*ABHD5*
- Neu–Laxova syndrome 1 [256520]		*PHGDH*
- Neu–Laxova syndrome 2 [616038]		*PSAT1*

AD autosomal dominant, *AR* autosomal recessive, *CDPX2* chondrodysplasia punctata type 2, *HMSN4* hereditary motor and sensory neuropathy type 4; OMIM in [ ], http://www.ncbi.nlm.nih.gov/omim

place regularly. The migration and differentiation of the cells from the stratum basale to the stratum granulosum takes place within about 14 days. It takes another 14 days for the corneocytes to exfoliate, resulting in a total epidermal turnover of approximately 28 days, which can be considerably shortened depending on the disease or skin condition.

The intermediate filaments of the epidermis, i.e., cytoskeletal structures of 10 nm, consist mainly of keratins. At least 54 functionally relevant keratin genes can be differentiated in terms of tissue and in the skin and its appendages (Table 3). Type I keratins differ from type II keratins in size (40–57.5 kDa and 53–67 kDa respectively) and charge (acidic or alkaline–neutral). Equimolar amounts of keratin types I and II polymerize with each other by spontaneous parallel arrangement to keratin dimers, which in turn polymerize by antiparallel arrangement to protofilaments. At the end electron density keratin filaments are formed from protofibrils with a diameter of 5–8 nm, which together form sound fibrils with a diameter of about 25 nm. The basal keratinocytes mainly produce keratin 14 and 5, as well as type IV collagen, laminins, and integrins. The basement membrane is thus a product of the epidermis.

In addition to keratins, lipids are of great importance for the structure of the epidermis. Besides the neutral lipids (glycerol phosphatides, triglycerides, free fatty acids), complex sphingolipids (ceramides and glycosylceramides) occur in the epidermis, which are otherwise only found in the nervous system. In the basal epidermis the phospholipids form the largest lipid fraction in the cell membrane. During the keratinization process, a considerable increase in neutral

may require referral for cardiac, audiologic, ophthalmologic, or dental evaluation.

A skin biopsy for histologic examination may provide additional diagnostic clues (Table 58.4). The molecular defect underlying PPKs may involve epidermal proteases as well as proteins of the keratinocyte cytoskeleton, cornified envelope, desmosome (see Fig. 56.8), or gap junction (Fig. 58.4, Table 58.5); the known underlying genetic mutations are summarized in Table 58.6[5,6]. Genetic analysis is sometimes helpful to establish the specific diagnosis, facilitate identification of affected family members, and enable prenatal diagnosis.

PRIMARY PALMOPLANTAR KERATODERMAS (PPKS)	
Disorder	**Additional features**
Hereditary PPKs	
Diffuse PPK, isolated	
Vörner–Unna–Thost	Epidermolytic
Nagashima, Kimonis	Non-epidermolytic
Bothnia	White/spongy upon water exposure; non-epidermolytic
"Greither"	Transgrediens & progrediens; variable pathology
Mal de Meleda (including Gamborg Nielsen)	Mutilating, frequent superinfections; non-epidermolytic
Diffuse PPK with associated features	
Hearing impairment:	
Vohwinkel syndrome	Mutilating, "honeycomb" PPK; starfish keratoses on knuckles
Bart–Pumphrey syndrome	Knuckle pads, leukonychia
PPK due to connexin 26 defect	"Honeycomb" PPK
Mitochondrial PPK	
High risk of acral SCC:*	
Huriez syndrome	Scleroatrophy
PPK with sex reversal	Female-to-male sex reversal
Hypotrichosis & nail dystrophy:	
Clouston syndrome	Pebbled/grid-like acral papules
Odonto-onycho-dermal dysplasia	Hypodontia, facial telangiectasias/erythema
Schöpf–Schulz–Passarge syndrome	Hidrocystomas, other adnexal neoplasms, hypodontia
Keratoderma–hypotrichosis–leukonychia totalis	Total leukonychia
Mutilating:*	
Olmsted syndrome	Periorificial/intertriginous keratotic plaques
Papillon–Lefèvre syndrome**	Periodontitis, pyogenic infections, psoriasiform plaques
Loricrin keratoderma	Mild generalized ichthyosis, "honeycomb" PPK
KLICK	Keratosis linearis, ichthyosis congenita, sclerosing keratoderma
Vohwinkel syndrome (see above)	
Mal de Meleda (see above)	
Cardiomyopathy:	
Naxos disease***	Woolly hair, arrhythmias in adolescence
Focal PPK, isolated	
Brünauer–Fuhs–Siemens, Wachters	Striate on palms, nummular on soles; see Table 58.6 for genetic subtypes
Focal PPK with associated features	
Pachyonychia congenita	Painful soles, dystrophic nails, oral leukokeratosis, steatocystomas/other cysts
Focal palmoplantar and gingival keratosis	Painful soles, gingival leukokeratosis; epidermolytic
Howel–Evans syndrome	Esophageal carcinoma
Richner–Hanhart syndrome (oculocutaneous tyrosinemia)	Dendritic keratitis, corneal ulcers, intellectual disability, painful keratosis
Carvajal syndrome***	Striate on palms; woolly hair, dilated cardiomyopathy; occasionally skin fragility, nail dystrophy, and hypodontia
PPK and woolly hair	Striate on palms; woolly hair, hypotrichosis, leukonychia

*Acral SCC can also develop in patients with mutilating PPK.
**_Haim–Munk syndrome_ is an allelic disorder that also features arachnodactyly, atrophic nail changes, and acro-osteolysis.
***The PPK in Naxos disease is occasionally focal/striate, overlapping with Carvajal syndrome.

Table 58.1 Primary palmoplantar keratodermas (PPKs). More common disorders are in **bold** font. Other genodermatoses that feature PPK are listed in Table 58.2. Of note, the pattern can be different on the hands and feet, e.g. diffuse on the feet and focal on the hands. Progrediens means progressive, referring to the tendency to involve additional sites (e.g. the elbows and knees) over time. Mutilating refers to the development of digital constrictions and possible autoamputation (pseudoainhum; see Table 58.3). SCC, squamous cell carcinoma.

Continued

PRIMARY PALMOPLANTAR KERATODERMAS (PPKS)	
Disorder	**Additional features**
Punctate PPK, isolated	
Buschke–Fischer–Brauer (type 1 punctate PPK) Spiny keratoderma (type 2 punctate PPK) Marginal papular keratoderma: acrokeratoelastoidosis, focal acral hyperkeratosis (type 3 punctate PPK) **Punctate keratoses of the palmar creases** Porokeratosis punctata palmaris et plantaris	
Punctate PPK with associated features	
Cole disease	Guttate hypopigmentation, calcinosis cutis
PLACK syndrome	*P*eeling skin, *l*eukonychia, *c*heilitis, *k*nuckle pads
Acquired PPKs	
Keratoderma climactericum **Aquagenic PPK** Pityriasis rubra pilaris Associated with hypothyroidism/myxedema Associated with cancer Associated with lymphedema Arsenical keratoses Drug-induced keratoderma (e.g. lithium, verapamil, venlafaxine, glucan, tegafur, capecitabine, imatinib, sorafenib, sunitinib) Induced by dioxin or halogenated herbicides Associated with dermatomyositis: pityriasis rubra pilaris-like (Wong) type, "mechanic's hands" in antisynthetase syndrome	

Table 58.1 Primary palmoplantar keratodermas (PPKs). (cont'd)

Fig. 58.2 Focal palmoplantar keratoderma. A Striate type with linear hyperkeratosis on the palm overlaying flexor tendons. **B** Areata type on the soles. *B, Courtesy, Alfons Krol, MD, and Dawn Siegel, MD.*

HEREDITARY KERATODERMAS

DIFFUSE PALMOPLANTAR KERATODERMA WITHOUT ASSOCIATED FEATURES (ISOLATED, NON-SYNDROMIC)

Diffuse Epidermolytic Palmoplantar Keratoderma (EPPK)

Synonyms: ■ Unna–Thost disease ■ Vörner disease ■ Vörner–Unna–Thost type PPK ■ Keratosis palmoplantaris diffusa Vörner–Unna–Thost

History

In 1880, Thost described a family with diffuse non-transgrediens PPK. This was followed by Unna's description of a clinically identical, autosomal dominant PPK in two families. Although "Unna–Thost type" has been used to designate non-epidermolytic PPK (NEPPK), in 1992, Küster et al.[7] reviewed the family described by Thost and found the histologic features of epidermolytic hyperkeratosis, which characterize the "Vörner type" of PPK. Subsequently, after keratin mutations were reported in epidermolytic PPK (EPPK), identical mutations were found in the descendants of the family studied by Thost[8]. This confusion underscores the need for careful histologic examination and sometimes multiple biopsy specimens to identify epidermolytic hyperkeratosis.

Epidemiology

EPPK is likely the most common form of diffuse keratoderma, with an estimated incidence of ≥4.4 per 100 000 in Northern Ireland[9].

Pathogenesis

Mutations in *KRT1* and *KRT9* underlie diffuse EPPK. The expression of *keratin 9* is limited to the suprabasal cell layers of palmar and plantar skin. In patients with diffuse EPPK, the majority of keratin 9

HERITABLE FORMS OF CUTIS LAXA AND RELATED CONDITIONS					
Disorder	**Inheritance**	**Gene**	**Protein**	**Distribution of cutis laxa**	**Additional features**
Heritable forms of cutis laxa					
ADCL	AD[†]	*ELN* *FBLN5*	Elastin Fibulin 5	Generalized	• Cardiac valve abnormalities, aortic dilatation (variable) • Emphysema (uncommon) • Hernias[‡]
ARCL type 1B ARCL type 1A	AR	*FBLN4 (EFEMP2)* *FBLN5*	Fibulin 4 Fibulin 5	Generalized	• Arterial or aortic tortuosity/aneurysms (1B) and stenosis • Emphysema (more severe in 1A) • Bladder diverticula, hernias[‡] • In 1B: joint laxity, arachnodactyly, fractures, micrognathia
ARCL type 1C (Urban–Rifkin–Davis syndrome)	AR	*LTBP4*	Latent TGF-β binding protein-4	Generalized	• Pulmonary malformations/ emphysema (severe, often fatal) • Pulmonary arterial stenosis, valvular defects • GI and GU malformations (diverticula, stenoses) • Micrognathia, large fontanelles • Joint laxity, hypotonia
ARCL type 2A (Debré type; allelic to wrinkly skin syndrome)	AR	*ATP6V0A2*	Vacuolar H^+-ATPase V0, subunit A2	Generalized; improves over time	• Translucent skin with prominent veins (especially 2B, DBS) • Ocular abnormalities: strabismus & high myopia (2A); corneal opacities & cataracts (DBS, PADCL) • Facial dysmorphism[§]; triangular face with more progeroid appearance (2B, DBS); large fontanelles • Microcephaly, partial pachygyria (2A), absent corpus callosum (2B) • Intellectual disability; neurologic degeneration (2A); dystonia/athetosis (DBS) • Congenital hip dysplasia, joint laxity, hypotonia, scoliosis, short stature; osteopenia (2B, DBS) • Cranial arterial tortuosity (DBS-A, PADCL) • O- and N-glycosylation defect (2A[‖])
ARCL type 2B De Barsy syndrome (DBS) B (ARCL type 3B)	AR	*PYCR1*	Pyrroline-5-carboxylate reductase 1	Generalized; may favor dorsal hands/feet (2B) or improve over time; occasionally fat pads (e.g. on buttocks; DBS-A)	
Progeroid ADCL (PADCL) De Barsy syndrome (DBS) A (ARCL type 3A)	AD AR	*ALDH18A1*	Pyrroline-5-carboxylate synthetase		
ARCL types 2C and 2D	AR	*ATP6V1E1* (2C) *ATP6V1A* (2D)	Vacuolar H^+-ATPase V1, subunits E1 and A	Generalized with sparse or subcutaneous fat; large skin folds on extremities, fat pads on buttocks; may improve over time (2D)	• Pneumothoraces • Cardiac abnormalities, aortic dilation • "Mask-like" triangular face, hypotonia; variable hip dysplasia, marfanoid habitus • Neurodevelopmental abnormalities (2D)
Gerodermia osteodysplastica (geroderma osteodysplasticum)	AR	*GORAB*	Golgin, RAB6-interacting	Primarily on dorsal hands/feet	• Malar hypoplasia, prognathism, oblique furrow lateral to eyebrows • Joint laxity • Osteoporosis, fractures, dwarfism
Macrocephaly, alopecia, CL, and scoliosis (MACS)	AR	*RIN2*	RAS and RAB interactor 2	Generalized, most pronounced on face	• Sparse hair • Gingival hypertrophy, facial dysmorphism • Macrocephaly • Scoliosis, joint laxity

Table 97.6 Heritable forms of cutis laxa (CL) and related conditions. In addition to ARCL types 2A and 2C, CL can occur in other congenital disorders of glycosylation, e.g. due to mutations in a component of oligomeric Golgi complex 7 (*COG7*; multiple-system malformations) or steroid 5-α-reductase 3 (*SRD5A3*; also ichthyosis, ocular colobomas, and midline brain malformations). Transaldolase deficiency can present with neonatal CL (generalized or of the forehead) that improves over time as well as hypertrichosis, hepatosplenomegaly, hemolytic anemia, and GU malformations. "Neonatal CL with a marfanoid phenotype" was described in an infant who lacked immunohistochemical evidence of laminin-β1 in the skin. AD, autosomal dominant; *ALDH18A1*, aldehyde dehydrogenase 18 family, member A1; AR, autosomal recessive; *EFEMP2*, EGF-containing fibulin-like extracellular matrix protein 2; GI, gastrointestinal; GU, genitourinary; TGF, transforming growth factor; X-R, X-linked recessive.

HERITABLE FORMS OF CUTIS LAXA AND RELATED CONDITIONS					
Disorder	**Inheritance**	**Gene**	**Protein**	**Distribution of cutis laxa**	**Additional features**
Heritable forms of cutis laxa					
Occipital horn syndrome	X-R	*ATP7A*	ATPase, Cu^{2+}-transporting, α-polypeptide	Generalized, most pronounced on face and neck	• Easy bruising, coarse hair (variable) • Arterial tortuosity • Chronic diarrhea; bladder diverticula • Long face, high forehead, hooked nose • Developmental delay • Wedge-shaped occipital calcifications • Joint laxity
Related heritable conditions that feature lax and/or redundant skin					
Arterial tortuosity syndrome	AR	*SLC2A10 (GLUT10)*	Solute carrier family 2, member 10 (a glucose transporter)	Generalized; soft, hyperextensible skin has also been described	• Telangiectasias on cheeks (variable) • Arterial tortuosity • Long face, micrognathia, high-arched palate • Joint laxity and contractures
Larsen-like syndrome	AR	*B3GAT3*	Glucuronosyltransferase I	Wrinkled acral skin	• Joint dislocations, craniofacial dysmorphism, short stature • Congenital heart defects
Costello syndrome	AR	*HRAS*	HRAS	Variable, especially on hands and feet	• Deep palmoplantar creases, acanthosis nigricans, periorificial papillomas • Curly or sparse hair, coarse facies • Cardiac malformations • Mental and growth retardation • Risk of malignancies, e.g. rhabdomyosarcoma
Donohue syndrome ("leprechaunism"; see Ch. 101)	AR	*INSR*	Insulin receptor	Variable	• Generalized lipodystrophy, hypertrichosis, acanthosis nigricans • Insulin resistance • Distinctive "elfin" facies • Severe growth retardation
Finnish-type amyloidosis	AD	*GSN*	Gelsolin	Generalized	• Corneal dystrophy • Polyneuropathy
Cranioectodermal dysplasia (Sensenbrenner syndrome)	AR	*IFT122* *IFT43*	Intraflagellar transport proteins 122 and 43	Generalized; redundant skin folds at neck, ankles, and wrists	• Sparse, fine hair; short, broad nails • Narrow thorax, short proximal limbs, joint laxity, brachydactyly, dolichocephaly, dysmorphic facies, dental anomalies • Nephronophthisis, hepatic fibrosis, retinal dystrophy
		WDR35 *WDR19*	WD repeat domain 35 and 19		
Circumferential skin creases, Kunze type	AD, AR	*MAPRE2*	Microtubule-associated protein RP/EB family member 2	Multiple circumferential creases/folds of skin on the extremities (see Fig. 97.12) > neck > trunk (excess rather than lax skin); may resolve over time	• Cleft palate, short stature, dysmorphic facies (e.g. epicanthal folds, small eyes), overfolded helices • Intellectual disability
	AD	*TUBB*	Tubulin-β class I		
Nonsyndromic "Michelin tire baby"	AD	?	?		• Affected skin may have excess fat or smooth muscle, and the latter is often associated with hypertrichosis; elastic fibers are usually normal (fragmentation noted in only one report)

†An AR form of cutis laxa due to a homozygous elastin mutation has been reported in two related families.
‡E.g. inguinal, umbilical, diaphragmatic.
§Features may include a broad forehead and midfacial hypoplasia as well as findings related to the lax skin (e.g. down-slanting palpebral fissures, long philtrum).
‖Can be assessed via analysis of the glycosylation of serum transferrin ± apolipoprotein C-III.

Table 97.6 Heritable forms of cutis laxa (CL) and related conditions. (cont'd)

particularly pulmonary complications, can cause considerable morbidity and mortality[64]. Association with congenital hemolytic anemia has also been reported[65]. The cutaneous and extracutaneous features of specific types of heritable CL are summarized in Table 97.6.

Generalized acquired CL without an identifiable trigger may be clinically indistinguishable from milder heritable forms of CL, e.g. late-onset autosomal dominant CL. Generalized or localized forms of acquired CL can also occur in association with a plasma cell dyscrasia (see Fig. 97.10B), as a consequence of drug intake (particularly penicillamine), and subsequent to a systemic or cutaneous inflammatory disorder (Table 97.7). In the latter situation, the inflammatory reaction is followed by progressive sagging of the skin, which can become exten-

- Mitral and aortic valve prolapse/dilatation and regurgitation
- Cystic medionecrosis of the aorta
- Neurologic features include macrocephaly, hydrocephalus, syringomyelia, and basilar invagination

- Variable prognosis depending on disease type and severity:
  - Types I and IV: normal life span
  - Type II: **death in perinatal period**
  - Type III: increased mortality in 3rd/4th decade due to **respiratory failure** (secondary to kyphoscoliosis) or head trauma

Ehlers-Danlos syndrome (EDS)

- Heterogeneous group characterized by abnormal collagen structure and/or function within the skin, joints, and vasculature (Table 4-13)
- Classical EDS (most common subtype)
  - May deliver/be delivered pre-term as a result of early rupture of fetal membranes; normal life span
  - Mucocutaneous manifestations
    - Velvety, soft, and doughy consistency of skin
    - **Marked hyperextensibility of skin**
    - **Poor wound healing ("cigarette paper" scars)**

Table 4-13. Ehlers-Danlos Syndrome Classification

Type	Genes	Inheritance	Skin Findings	Joint Changes	Other
Classical (formerly **Type I**, "gravis" and **Type II**, "mitis")	***COL5A1***, ***COL5A2***	AD	Hyperextensibility Easy bruising Fragile skin Widened atrophic ("**fish mouth**" or "**cigarette paper**") scars **Molluscoid pseudotumors** (overlying extensor joints and pressure points) Spheroids	Hypermobility and joint dislocations	**(+)Gorlin's sign** Absence of lingual frenulum
Hypermobility (formerly **Type III**)	*TNXB*	AD/AR	Mild	**Hypermobility** Chronic joint pain + arthritis Recurrent **dislocations and subluxations**	Deficiency of **tenascin X**
Vascular (formerly **Type IV**)	***COL3A1***	AD	Thin, translucent skin Extensive bruising Early varicosities (can **visualize veins under skin** easily) Acrogeria	Small joint hypermobility	Mnemonic: "IV = blood vessels" **Rupture of bowel, uterus, or arteries** Most **life-threatening** form
Kyphoscoliotic (formerly **Type VI**)	Lysyl hydroxylase 1 (*PLOD1*)	AR	Mild	Hypermobility **Severe scoliosis**	Severe muscle hypotonia **Ruptured globe, blindness, retinal detachment, or keratoconus** Marfanoid Osteopenia Ascorbic acid supplementation may help
Arthrochalasic (formerly **Types VIIa and VIIb**)	*COL1A1*, *COL1A2*	AD	Mild	**Most severe hypermobility** with recurrent subluxations/ dislocations (much more severe than Hypermobility type)	**Congenital hip dislocation** Short stature
Dermatosparaxis (formerly **Type VIIc**)	Procollagen N-proteinase (***ADAMTS2***)	AR	Severe fragility **Sagging**, **redundant skin**, and bruising	Mild	Umbilical/inguinal hernias Premature rupture of fetal membranes
Periodontitis type (formerly **Type VIII**)	Unclear	AD	Hyperextensible skin with scarring (esp. pretibial) and bruising	Mild	**Severe periodontitis → teeth loss**

(Adapted from Paller S, Mancini AJ. Hurwitz Clinical Pediatric Dermatology, 4th Ed. Elsevier. 2011)

Insect repellents containing diethyltoluamide (DEET) offer the most effective protection against mosquitoes, flies, fleas, mites, and ticks. Ethyl hexanediol, dimethyl phthalate, and dimethyl carbate butopyronoxyl provide a narrower spectrum of prevention compared with DEET. Use of a combination of these repellants improves effectiveness. Repellants form a barrier against penetration that extends <4 cm away from the skin. DEET also blocks the mosquitoes ability to track human carbon dioxide vapor trails. Repellants do not mask other insect stimuli such as sweat, moisture, and warmth. DEET protection lasts about 10 to 12 hours at room temperature, but can be shortened by clothing friction, water, sweat, wind, or heat. Repellants may last several days on fabrics. Mosquito nets and clothing should also be used in areas where mosquitoes serve as vectors for disease.

MITES

I. BACKGROUND Mites are a form of arachnid that parasitize humans and feed on organic matter. Depending on the type of mite, a range of dermatologic manifestations are seen.

II. CLINICAL PRESENTATION Mite infestations manifest as papulosquamous, urticarial, inflammatory, or even bullous dermatoses. They are also responsible for transmitting infectious diseases. Clinical manifestations and diseases associated with specific mites are outlined in Table 6-6. Cheyletiella mites eat keratin on small mammals, giving dogs and cats "walking dandruff."

TABLE 6-6 Mite-Associated Disease

Mite	Associated Disease	Treatment
Acarus (grain mite)	Baker itch	Pest control Symptomatic relief[a]
Allodermanyssus sanguineus (house mouse mite)	Rickettsial pox	Doxycycline[b]
Cheyletiella	Walking dandruff	Treat pets: - Ectoparasitic shampoos and dips) - Permethrin spray - Fipronil spray - Topical amitraz Symptomatic relief[a]
Demodex	Rosacea	Topical sulfur (initial treatment) Other topicals: - Permethrin 5% - Salicylic acid

(*Continued*)

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TABLE 6-6 Mite-Associated Disease (Continued)

		- Metronidazole - Crotamiton - Lindane - Retinoids Oral ivermectin Oral retinoids
Dermanyssus (fowl mite) *Ornithonyssus* (fowl mite)	Equine encephalitis Western equine encephalitis	Removal of infested birds and nests House cleaning and fumigation Symptomatic relief[a] Topical acaricides - Gamma benzene hexachloride 1% - Crotamiton - Permethrin - Malathion
Dermatophagoides (dust mite)	Allergic reactions Atopic dermatitis	Humidity < 50% Mattress and pillow protective coverings Wash bedding once weekly in hot water Symptomatic relief[a]
Glycyphagus (cheese mite)	Grocer itch	Pest control Symptomatic relief[a]
Sarcoptes scabei (scabies mite)	Pruritic dermatitis	Wash clothes and bedding in hot water Pest control Topical permethrin Oral ivermectin
Trombicula (chigger mite)	Dermatitis and scrub typhus	Symptomatic relief,[a] additionally: - Antipruritics: camphor, menthol - Topical anesthetics with pramoxine - Potent topical corticosteroids under occlusion - Ice application - Excision of refractory lesions

[a]Symptomatic relief for mite-associated dermatitis includes topical corticosteroids and oral antihistamines. [b]Antibiotics may shorten the duration of symptoms. Second-line antibiotics include erythromycin, tetracycline, and chloramphenicol.

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Table 12.1 Primary human demodicosis: proposed classification and nomenclature

Proposed nomenclature	Definition and clinical manifestations
Spinulate demodicosis	Discrete fine, whitish to yellowish, spiky, changes involving sebaceous hair follicles, with or without faint erythema and little inflammation
Papulopustular demodicosis, perioral demodicosis, periorbital demodicosis, periauricular demodicosis	Papulopustules mostly involving the face (predilection for perioral, periorbital, and periauricular regions), in patients without (primary form) or with preexisting inflammatory dermatoses (secondary form)
Nodulocystic/ conglobate demodicosis	Intense immune reaction with massive follicular and perifollicular inflammatory infiltrates caused by *Demodex* proliferation, pus accumulation, and suppurative succulent changes

Adapted from Chen et al. [16]

Dermatology diseases Index

FIRST INDEX OF SKIN DISEASES IN THE WORLD

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& Leprosy Hospital, Egypt

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